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RECORDS LAST ADDED: 9 July 2003 (20030709/ED)
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L88 ANSWER 1 OF 4 BIOSIS COPYRIGHT 2003 BIOLOGICAL ABSTRACTS INC.
     1996:556811 BIOSIS
DN
     PREV199699279167
TI
     Quality control of DNA sequencing templates and contig assembly
     using quantitative DNA fiber mapping (QDFM) facilities parallelism in
     producing sequencing.
     Wang, M. (1); Ericsson, C.; Martin, C.; Collins, C. (1);
ΑU
     Palazzolo, M.; Gray, J. W. (1); Weier, H.-U. G. (1)
CS
     (1) Resource Mol. Cytogenet., Life Sci. Div., Univ. California, Lawrence
     Berkeley Natl. Lab., Berkeley, CA 94720 USA
SO
    American Journal of Human Genetics, (1996) Vol. 59, No. 4 SUPPL., pp.
    Meeting Info.: 46th Annual Meeting of the American Society of Human
```

ISSN: 0002-9297. DTConference

LA English

CC General Biology - Symposia, Transactions and Proceedings of Conferences, Congresses, Review Annuals 00520 General Biology - Information, Documentation, Retrieval and Computer Applications 00530 Genetics and Cytogenetics - Human *03508 Mathematical Biology and Statistical Methods *04500 Biochemical Methods - Nucleic Acids, Purines and Pyrimidines 10052

Genetics San Francisco, California, USA October 29-November 2, 1996

Biochemical Studies - Nucleic Acids, Purines and Pyrimidines

Replication, Transcription, Translation *10300 Biophysics - Molecular Properties and Macromolecules *10506 Metabolism - Nucleic Acids, Purines and Pyrimidines *13014

BCHominidae *86215

ITMajor Concepts

> Biochemistry and Molecular Biophysics; Genetics; Mathematical Biology (Computational Biology); Metabolism; Molecular Genetics (Biochemistry and Molecular Biophysics)

IT Miscellaneous Descriptors

GENE EXPRESSION; HUMAN GENOME; MARKERS; MEETING ABSTRACT; MEETING POSTER; MOLECULAR GENETICS

ORGN Super Taxa

Hominidae: Primates, Mammalia, Vertebrata, Chordata, Animalia

ORGN Organism Name

Hominidae (Hominidae)

ORGN Organism Superterms

animals; chordates; humans; mammals; primates; vertebrates

T.88 ANSWER 2 OF 4 BIOSIS COPYRIGHT 2003 BIOLOGICAL ABSTRACTS INC.

1996:100562 BIOSIS ΑN

DN PREV199698672697

A statistical method for the analysis of comparative genomic hybridization TI

their own but you

profiles. Mohapatra, Gavatry (1); Moore, Dan (1); Sudar, Damir (1); Pinkel, Dan (1); ΑU Gray, Joe (1); Feuerstein, Burt CS (1) Div. Mol. Cytometry, Dep. Lab. Med., UCSF, San Francisco, CA 94143 USA SO Cancer Genetics and Cytogenetics, (1995) Vol. 84, No. 2, pp. 151. Meeting Info.: Sixth International Workshop on Chromosomes in Solid Tumors Tucson, Arizona, USA February 19-21, 1995 ISSN: 0165-4608. DTConference LA English CC General Biology - Symposia, Transactions and Proceedings of Conferences, Congresses, Review Annuals Cytology and Cytochemistry - Human *02508 Genetics and Cytogenetics - Human *03508 Mathematical Biology and Statistical Methods $\star 04500$ Biochemical Studies - Nucleic Acids, Purines and Pyrimidines Neoplasms and Neoplastic Agents - Biochemistry *24006 Neoplasms and Neoplastic Agents - Carcinogens and Carcinogenesis *24007 BC Hominidae *86215 ΙT Major Concepts Biochemistry and Molecular Biophysics; Cell Biology; Genetics; Mathematical Biology (Computational Biology); Oncology (Human Medicine, Medical Sciences) Miscellaneous Descriptors CANCER GENETICS; DNA COPY NUMBER; MEETING ABSTRACT; STATISTICAL METHOD ORGN Super Taxa Hominidae: Primates, Mammalia, Vertebrata, Chordata, Animalia ORGN Organism Name human (Hominidae) ORGN Organism Superterms animals; chordates; humans; mammals; primates; vertebrates ANSWER 3 OF 4 BIOSIS COPYRIGHT 2003 BIOLOGICAL ABSTRACTS INC. L88 ΑN 1995:528155 BIOSIS DN PREV199598542455 TIQuantitative DNA fiber mapping. Weier, H.-U. G. (1); Wang, M.; Mullikin, J. C.; Zhu, Y.; Cheng, J.-F.; ΑIJ Greulich, K. M.; Bensimon, A; Gray, J. W. CS (1) Cent. Molecular Cytogenetics, Life Sci. Div., MS 74-157, Univ. California, Lawrence Berkeley Lab., 1 Cyclotron Rd., Berkeley, CA 94720 SO Human Molecular Genetics, (1995) Vol. 4, No. 10, pp. 1903-1910. ISSN: 0964-6906. DTArticle LA English AΒ The assembly of sequence ready, high-resolution physical maps and construction of minimally overlapping contigs for the human as well as model genomes requires accurate determination of the extent of overlap between adjacent clones as well as their relative orientation. This is presently done by procedures such as clone fingerprinting, Southern blot analysis or clone end sequencing. We present a complementary analytical technique to map directly cloned DNA sequences on to individual stretched DNA molecules. This approach uses the hydrodynamic

to individual stretched DNA molecules. This approach uses the hydrodynamic force of a receding meniscus to prepare straight high molecular weight DNA molecules that provide a linear template of apprx 2.3 kb/mu-m on to which the cloned probes can be mapped by in situ hybridization. This technique has numerous advantages such as a very high density of mapping templates, reproducible stretching of the mapping template providing a linear genomic scale, determination of clone orientation and direct visualization of DNA repeats. The utility and accuracy of quantitative DNA fiber mapping are illustrated through three examples: (i) mapping of lambda DNA restriction

fragments along linearized apprx 49 kb long lambda phage DNA molecules with apprx 1 kb precision; (ii) localization of the overlap between a cosmid and a colinear P1 clone; and (iii) mapping of P1 clones along a apprx 490 kb yeast artificial chromosome (YAC) with apprx 5 kb precision and estimation of the apprx 25 kb gap between them. Methods, Materials and Apparatus, General - Photography *01012 Cytology and Cytochemistry - Plant *02504 Genetics and Cytogenetics - Plant *03504 Radiation - Radiation and Isotope Techniques *06504 Biochemical Methods - Nucleic Acids, Purines and Pyrimidines *10052 Biochemical Studies - Nucleic Acids, Purines and Pyrimidines *10062 Biophysics - General Biophysical Techniques *10504 Biophysics - Molecular Properties and Macromolecules *10506 External Effects - Pressure *10606 Genetics of Bacteria and Viruses *31500 Virology - Bacteriophage *33504 BC Siphoviridae 02710 Fungi - Unspecified *15000 ΙT Major Concepts Biochemistry and Molecular Biophysics; Cell Biology; Equipment, Apparatus, Devices and Instruments; Genetics; Methods and Techniques; Microbiology; Physiology; Radiology (Medical Sciences) Miscellaneous Descriptors ΙT FLUORESCENCE IN-SITU HYBRIDIZATION; HYDRODYNAMIC FORCE; LAMBDA DNA RESTRICTION FRAGMENTS; QUANTITATIVE IMAGE ANALYSIS; YEAST ARTIFICIAL CHROMOSOME ORGN Super Taxa Fungi - Unspecified: Fungi, Plantae; Siphoviridae: Viruses ORGN Organism Name fungi (Fungi - Unspecified); Siphoviridae (Siphoviridae) ORGN Organism Superterms fungi; microorganisms; nonvascular plants; plants; viruses ANSWER 4 OF 4 BIOSIS COPYRIGHT 2003 BIOLOGICAL ABSTRACTS INC. ΑN 1995:476706 BIOSIS PREV199598491006 Statistical method for testing genetic gain or loss in CGH profiles. Magrane, G. G.; Moore, D. H. Ii; Cronin, J. E.; Yu, L. C.; Pinkel, D.; DMC/MCB 230, Box 0808, Univ. Calif., San Francisco, CA 94103-0808 USA American Journal of Human Genetics, (1995) Vol. 57, No. 4 SUPPL., pp. A72. Meeting Info.: 45th Annual Meeting of the American Society of Human Genetics Minneapolis, Minnesota, USA October 24-28, 1995 Their buy ISSN: 0002-9297. Conference LA . English General Biology - Symposia, Transactions and Proceedings of Conferences, Congresses, Review Annuals Cytology and Cytochemistry - Human *02508 Genetics and Cytogenetics - Human *03508 Mathematical Biology and Statistical Methods Biochemical Studies - Nucleic Acids, Purines and Pyrimidines Neoplasms and Neoplastic Agents - General *24002 Neoplasms and Neoplastic Agents - Carcinogens and Carcinogenesis *24007 Hominidae *86215 Major Concepts Cell Biology; Genetics; Oncology (Human Medicine, Medical Sciences) Miscellaneous Descriptors CHROMOSOME; COMPARATIVE GENOMIC HYBRIDIZATION; DNA; MEETING

DN

TΤ

ΑIJ

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BC

ΙT

IT

ABSTRACT; MEETING POSTER

ORGN Super Taxa

Hominidae: Primates, Mammalia, Vertebrata, Chordata, Animalia

ORGN Organism Name

human (Hominidae)

ORGN Organism Superterms

animals; chordates; humans; mammals; primates; vertebrates

=> d all

L90 ANSWER 1 OF 1 BIOSIS COPYRIGHT 2003 BIOLOGICAL ABSTRACTS INC.

AN **1987:414134** BIOSIS

DN BR33:83812

TI AUTOMATED QUANTIFICATION OF THE FREQUENCY OF ABERRANT CHROMOSOMES IN HUMAN CELLS.

AU LUCAS J; LOZES C; MULLIKIN J; PINKEL D; GRAY J

CS LAWRENCE LIVERMORE NATL. LAB., LIVERMORE, CALIF. 94550.

SO XIITH INTERNATIONAL MEETING OF THE SOCIETY FOR ANALYTICAL CYTOLOGY, CAMBRIDGE, ENGLAND, UK, AUGUST 9-15, 1987. CYTOMETRY. (1987) 0 (SUPPL 1), 13.

CODEN: CYTODQ. ISSN: 0196-4763.

DT Conference

FS BR; OLD

LA English

General Biology - Symposia, Transactions and Proceedings of Conferences, Congresses, Review Annuals 00520
Cytology and Cytochemistry - Human *02508

Genetics and Cytogenetics - Human *03508

Mathematical Biology and Statistical Methods *04500

Radiation - Radiation Effects and Protective Measures *06506 Blood, Blood-Forming Organs and Body Fluids - Blood, Lymphatic and Reticuloendothelial Pathologies *15006 Blood, Blood-Forming Organs and Body Fluids - Lymphatic Tissue and

Reticuloendothelial System *15008

BC Hominidae 86215

IT Miscellaneous Descriptors

ABSTRACT LYMPHOBLASTOID CELLS GAMMA-RAYS

=> d 192 all tot

L92 ANSWER 1 OF 9 BIOSIS COPYRIGHT 2003 BIOLOGICAL ABSTRACTS INC.

AN 2002:386802 BIOSIS

DN PREV200200386802

TI Genomic copy number changes in mouse primary breast tumors and their metastases identified using comparative genomic hybridization to DNA microarrays.

AU Hodgson, Graeme (1); Tirone, Jamie; Malek, Tiffany; Hariono, Sujatmi; Pinkel, Daniel; Albertson, Donna G.; Muller, William J.; Gray, Joe W.

CS (1) University of California, San Francisco, San Francisco, CA USA SO Proceedings of the American Association for Cancer Research Annual

Meeting, (March, 2002) Vol. 43, pp. 292. print.
Meeting Info.: 93rd Annual Meeting of the American Association for Cancer Research San Francisco, California, USA April 06-10, 2002 ISSN: 0197-016X.

DT Conference

LA English

CC General Biology - Symposia, Transactions and Proceedings of Conferences, Congresses, Review Annuals *00520
Cytology and Cytochemistry - Animal *02506
Genetics and Cytogenetics - General *03502
Genetics and Cytogenetics - Animal *03506

borin - 09 / 586529 Biochemical Studies - Nucleic Acids, Purines and Pyrimidines Respiratory System - Physiology and Biochemistry *16004 Reproductive System - Physiology and Biochemistry *16504 Reproductive System - Pathology *16506 Neoplasms and Neoplastic Agents - Pathology; Clinical Aspects; Systemic Effects *24004 Muridae 86375 Major Concepts Methods and Techniques; Molecular Genetics (Biochemistry and Molecular Biophysics); Tumor Biology Parts, Structures, & Systems of Organisms breast: reproductive system; chromosome 1: locus p32-36; chromosome 15; chromosome 19; chromosome 2; chromosome 4; chromosome 6; lung: respiratory system; mammary epithelial cell: reproductive system Diseases genomic copy number abnormality: genetic disease; primary breast tumor: neoplastic disease, reproductive system disease/female Chemicals & Biochemicals ErbB-2: activation, expression, mutation, phosphorylation; NBL-1; genome; k-Ras; tumor necrosis factor receptor Methods & Equipment DNA microarray: analytical method; comparative genomic hybridization: analytical method Miscellaneous Descriptors Meeting Abstract ORGN Super Taxa Muridae: Rodentia, Mammalia, Vertebrata, Chordata, Animalia ORGN Organism Name mouse (Muridae) ORGN Organism Superterms Animals; Chordates; Mammals; Nonhuman Mammals; Nonhuman Vertebrates; Rodents; Vertebrates L92 ANSWER 2 OF 9 BIOSIS COPYRIGHT 2003 BIOLOGICAL ABSTRACTS INC. **2002:159019** BIOSIS PREV200200159019 High-resolution assessment of DNA loss in RIP-Tag mouse pancreatic tumors using comparative genomic hybridization to DNA microarrays. Hodgson, Graeme (1); Hager, Jeff; Collins, Colin; Wernick, Meredith; Werhane, Heather; Albertson, Donna; Pinkel, Daniel; Hanahan, Douglas; Gray, Joe (1) UCSF Cancer Center, San Francisco, CA USA Cytometry Supplement, (2000) No. 10, pp. 179. print. Meeting Info.: The XX Congress of the International Society for Analytical Cytology Montpellier, France May 20-25, 2000 International Society for Analytical Cytology . ISSN: 1046-7386. Conference English General Biology - Symposia, Transactions and Proceedings of Conferences, Congresses, Review Annuals *00520 Genetics and Cytogenetics - General *03502 Genetics and Cytogenetics - Animal *03506 Biochemical Studies - Nucleic Acids, Purines and Pyrimidines *10062 Endocrine System - Pancreas *17008 Neoplasms and Neoplastic Agents - Pathology; Clinical Aspects; Systemic Effects *24004

BCMuridae 86375 ΙT Major Concepts

BC.

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DT

LA

Methods and Techniques; Molecular Genetics (Biochemistry and Molecular Biophysics); Tumor Biology

```
ΙT
     Parts, Structures, & Systems of Organisms
        chromosome-16; chromosome-9
IT
     Diseases
        insulinoma: endocrine disease/pancreas, neoplastic disease
TT
     Chemicals & Biochemicals
        DNA
TΤ
     Alternate Indexing
        Insulinoma (MeSH)
ΙŢ
     Methods & Equipment
        comparative genomic hybridization: genetic method; comparative genomic
        hybridization to DNA microarray: genetic method
TΤ
     Miscellaneous Descriptors
        loss of heterozygosity; Meeting Abstract
ORGN Super Taxa
        Muridae: Rodentia, Mammalia, Vertebrata, Chordata, Animalia
ORGN Organism Name
        mouse (Muridae): RIP-Tag
ORGN Organism Superterms
        Animals; Chordates; Mammals; Nonhuman Mammals; Nonhuman Vertebrates;
        Rodents; Vertebrates
L92 ANSWER 3 OF 9 BIOSIS COPYRIGHT 2003 BIOLOGICAL ABSTRACTS INC.
     2002:158601 BIOSIS
DN
     PREV200200158601
TI
     High-resolution assessment of DNA loss in RIP-Tag mouse pancreatic tumors
     using comparative genomic hybridization to DNA microarrays.
     Hodgson, Graeme (1); Hager, Jeff (1); Collins, Colin (1);
ΑU
     Wernick, Meredith (1); Werhane, Heather (1); Pinkel, Daniel (1); Hanahan,
     Douglas (1); Gray, Joe (1); Albertson, Donna
     (1) University of California Cancer Center, San Francisco, CA USA
CS
SO
     Cytometry Supplement, (2000) No. 10, pp. 60. print.
     Meeting Info.: The XX Congress of the International Society for
     Analytical Cytology Montpellier, France May 20-25, 2000 International
     Society for Analytical Cytology
     . ISSN: 1046-7386.
DT
     Conference
LA
     English
CC
     General Biology - Symposia, Transactions and Proceedings of
     Conferences, Congresses, Review Annuals *00520
     Genetics and Cytogenetics - General *03502
     Genetics and Cytogenetics - Animal *03506
     Biochemical Studies - General *10060
       Biochemical Studies - Nucleic Acids, Purines and Pyrimidines
     *10062
     Endocrine System - General *17002
     Endocrine System - Pancreas *17008
     Neoplasms and Neoplastic Agents - Pathology; Clinical Aspects; Systemic
     Effects *24004
BC
    Muridae
               86375
IT
    Major Concepts
        Biochemistry and Molecular Biophysics; Genetics; Methods and
        Techniques; Tumor Biology
ΙT
     Parts, Structures, & Systems of Organisms
        chromosome 16: deletion; chromosome 9: deletion; pancreatic islet beta
        cell: endocrine system
ΙT
     Diseases
        insulinoma: endocrine disease/pancreas, neoplastic disease
ΤТ
    Chemicals & Biochemicals
        DNA: copy number, high-resolution assessment, loss, microarray; SV40
        large T-antigen
IT
    Alternate Indexing
        Insulinoma (MeSH)
```

ΙT

Methods & Equipment

```
comparative genomic hybridization: assessment method
ΙT
     Miscellaneous Descriptors
          Meeting Abstract
ORGN Super Taxa
        Muridae: Rodentia, Mammalia, Vertebrata, Chordata, Animalia
ORGN Organism Name
        mouse (Muridae): RIP-Tag, model, transgenic
ORGN Organism Superterms
        Animals; Chordates; Mammals; Nonhuman Mammals; Nonhuman Vertebrates;
        Rodents; Vertebrates
     ANSWER 4 OF 9 BIOSIS COPYRIGHT 2003 BIOLOGICAL ABSTRACTS INC.
AN
     2002:158568 BIOSIS
DN
     PREV200200158568
     Oligonucleotide-array-based comparative genomic hybridization.
TI
ΑU
     Baldocchi, Russ (1); Kowbel, Dave (1); Collins, Colin (1);
     Glynne, Richard; Tom, Ed; Mack, David; Gray, Joe
CS
     (1) Cancer Center, University of California at San Francisco, San
     Francisco, CA USA
SO
     Cytometry Supplement, (2000) No. 10, pp. 50. print.
     Meeting Info.: The XX Congress of the International Society for Analytical Cytology Montpellier, France May 20-25, 2000 International
     Society for Analytical Cytology
     . ISSN: 1046-7386.
DT
     Conference
LA
     English
CC.
     General Biology - Symposia, Transactions and Proceedings of
     Conferences, Congresses, Review Annuals *00520
     Genetics and Cytogenetics - General *03502
       Biochemical Studies - Nucleic Acids, Purines and Pyrimidines
     *10062
     Reproductive System - Pathology *16506
     Neoplasms and Neoplastic Agents - Pathology; Clinical Aspects; Systemic
     Effects *24004
     Major Concepts
ΙT
        Genetics; Methods and Techniques; Tumor Biology
IT
     Diseases
        breast cancer: neoplastic disease, reproductive system disease/female;
        ovarian cancer: neoplastic disease, reproductive system disease/female
     Chemicals & Biochemicals
IΤ
        DNA; oligonucleotide
IT
     Alternate Indexing
        Breast Neoplasms (MeSH); Ovarian Neoplasms (MeSH)
IT
     Methods & Equipment
        PCR [polymerase chain reaction]: DNA amplification, amplification
        method, in situ recombinant gene expression detection, sequencing
        techniques; oligonucleotide-array-based comparative genomic
        hybridization: assessment method
ΙT
     Miscellaneous Descriptors
        genomic copy number: abnormality; Meeting Abstract
L92
     ANSWER 5 OF 9 BIOSIS COPYRIGHT 2003 BIOLOGICAL ABSTRACTS INC.
AN
     2002:158546 BIOSIS
DN
     PREV200200158546
TI
     Technical approaches for producing and analyzing DNA microarrays.
     Hamilton, Greg (1); Gray, Joe (1); Albertson, Donna (1); Pinkel,
ΑU
     Dan (1); Jones, Arthur; Uber, Don; Davy, Donn; Hansen, Tony; Nordmeyer,
     Robert; Wilson, Dave; Jaklevic, Joe
CS
     (1) UCSF Cancer Center, San Francisco, CA USA
     Cytometry Supplement, (2000) No. 10, pp. 43. print.
SO
     Meeting Info.: The XX Congress of the International Society for
     Analytical Cytology Montpellier, France May 20-25, 2000 International
```

Society for Analytical Cytology

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FILE COVERS 1907 - 15 Jul 2003 VOL 139 ISS 3 FILE LAST UPDATED: 14 Jul 2003 (20030714/ED)

This file contains CAS Registry Numbers for easy and accurate substance identification.

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L117 ANSWER 1 OF 19 HCAPLUS COPYRIGHT 2003 ACS

AN 2003:524409 HCAPLUS

TI End-sequence profiling: Sequence-based analysis of aberrant genomes

AU Volik, Stanislav; Zhao, Shaying; Chin, Koei; Brebner, John H.; Herndon,
David R.; Tao, Quanzhou; Kowbel, David; Huang, Guiqing; Lapuk, Anna; Kuo,
Wen-Lin; Magrane, Gregg; de Jong, Pieter; Gray, Joe W.; Collins, Colin

CS Cancer Research Institute, University of California Comprehensive Cancer Center, San Francisco, CA, 94115, USA

SO Proceedings of the National Academy of Sciences of the United States of America (2003), 100(13), 7696-7701 CODEN: PNASA6; ISSN: 0027-8424

PB National Academy of Sciences

DT Journal

LA English

CC 3-3 (Biochemical Genetics)
Section cross-reference(s): 13, 14

ΑB Genome rearrangements are important in evolution, cancer, and other diseases. Precise mapping of the rearrangements is essential for identification of the involved genes, and many techniques have been developed for this purpose. End-sequence profiling (ESP) is now shown to be particularly well suited to this purpose. ESP is accomplished by constructing a bacterial artificial chromosome (BAC) library from a test genome, measuring BAC end sequences, and mapping end-sequence pairs onto the normal genome sequence. Plots of BAC end-sequences d. identify copy no. abnormalities at high resoln. BACs spanning structural aberrations have end pairs that map abnormally far apart on the normal genome sequence. These pairs can then be sequenced to det. the involved genes and breakpoint sequences. ESP anal. of the breast cancer cell line MCF-7 demonstrated its utility for anal. of complex genomes. End sequencing of .apprx.8000 clones (0.37-fold haploid genome clonal coverage) produced a comprehensive genome copy no. map of the MCF-7 genome at better than 300-kb resoln. and identified 381 genome breakpoints, a subset of which was verified by fluorescence in situ hybridization mapping and sequencing. The sequences reported in this paper have been deposited in the GenBank/EMBL/DDBJ database with accession nos. BZ597614-BZ612944 and [This abstr. record is one of four records for this document necessitated by the large no. of index entries required to fully index the document and publication system constraints.].

ST end sequence profiling aberrant genome analysis; breast cancer MCF7 genome analysis end sequence profiling

IT Genomic library

(BAC; end-sequence profiling for sequence-based anal. of aberrant genomes as applied to the breast cancer cell line MCF-7)

IT Genetic methods

(ESP (end-sequence profiling); end-sequence profiling for sequence-based anal. of aberrant genomes as applied to the breast cancer cell line MCF-7)

IT Animal cell line

```
530774-48-2, GenBank BZ600980
                                      530774-49-3, GenBank BZ600981
     530774-50-6, GenBank BZ600982
                                      530774-51-7, GenBank BZ600983
     530774-52-8, GenBank BZ600984
                                      530774-53-9, GenBank BZ600985
     530774-54-0, GenBank BZ600986
                                      530774-55-1, GenBank BZ600987
     530774-56-2, GenBank BZ600988
530774-58-4, GenBank BZ600990
                                      530774-57-3, GenBank BZ600989
                                      530774-59-5, GenBank BZ600991
     530774-60-8, GenBank BZ600992
530774-62-0, GenBank BZ600994
                                      530774-61-9, GenBank BZ600993
                                      530774-63-1, GenBank BZ600995
     530774-64-2, GenBank BZ600996
                                      530774-65-3, GenBank BZ600997
     530774-66-4, GenBank BZ600998
                                      530774-67-5, GenBank BZ600999
     530774-68-6, GenBank BZ601000
     RL: BSU (Biological study, unclassified); PRP (Properties); BIOL
     (Biological study)
        (nucleotide sequence; end-sequence profiling for sequence-based anal.
        of aberrant genomes as applied to the breast cancer cell line MCF-7)
L117 ANSWER 5 OF 19 HCAPLUS COPYRIGHT 2003 ACS
     2002:755055 HCAPLUS
     137:258485
    Methods for diagnosing and monitoring ovarian cancer by profiling
     associated marker genes using comparative genomic hybridization array.
     Chin, Koei; Kuo, Wen-lin; Pinkel, Daniel; Albertson, Donna; Collins,
     Colin; Gray, Joe W.
     USA
     U.S. Pat. Appl. Publ., 24 pp.
     CODEN: USXXCO
     Patent
     English
     ICM C12Q001-68
     435006000
     3-1 (Biochemical Genetics)
     Section cross-reference(s): 14
FAN.CNT 1
     PATENT NO.
                      KIND DATE
                                            APPLICATION NO.
                                                             DATE
     US 2002142305
                       A1 ·
                            20021003
                                            US 2001-819103
                                                             20010327
    WO 2002077292
                       Α1
                            20021003
                                           WO 2002-US9804
                                                             20020326
         W: AE, AG, AL, AM, AT, AU, AZ, BA, BB, BG, BR, BY, BZ, CA, CH, CN,
             CO, CR, CU, CZ, DE, DK, DM, DZ, EC, EE, ES, FI, GB, GD, GE, GH,
             GM, HR, HU, ID, IL, IN, IS, JP, KE, KG, KP, KR, KZ, LC, LK, LR,
             LS, LT, LU, LV, MA, MD, MG, MK, MN, MW, MX, MZ, NO, NZ, OM, PH,
             PL, PT, RO, RU, SD, SE, SG, SI, SK, SL, TJ, TM, TN, TR, TT, TZ,
             UA, UG, UZ, VN, YU, ZA, ZM, ZW, AM, AZ, BY, KG, KZ, MD, RU, TJ, TM
         RW: GH, GM, KE, LS, MW, MZ, SD, SL, SZ, TZ, UG, ZM, ZW, AT, BE, CH,
             CY, DE, DK, ES, FI, FR, GB, GR, IE, IT, LU, MC, NL, PT, SE, TR,
             BF, BJ, CF, CG, CI, CM, GA, GN, GQ, GW, ML, MR, NE, SN, TD, TG
PRAI US 2001-819103
                            20010327
    This invention pertains to the discovery that an amplification of some
    genes or an increase in that gene activity and a deletion of some genes or
    a decrease in that gene activity is a marker for the presence of,
    progression of, or predisposition to, a cancer (e.g., ovarian cancer).
    Using this information, this invention provides methods of detecting a
    predisposition to cancer in an animal. The methods involve (i) providing
    a biol. sample from an animal (e.g. a human patient); (ii) detecting the
    level of the genes of the present invention within the biol. sample; and
    (iii) comparing the level of one or more of said genes with a level of one
    or more of said genes in a control sample taken from a normal, cancer-free
    tissue. In particular, array comparative genomic hybridization using
    5'-amino-linked degenerate oligonucleotide primer (DOP) PCR is used to
    analyze gene amplification or deletion assocd. ovarian cancer. Approx.
    twenty amplified or deleted (with >0.4% fold gain or loss of expression at
    mRNA level) genomic regions with ref. GenBank nos. are identified to be
    assocd. with human ovarian tumors. Gene-specific arrays targeted to these
```

ΑN DN

IN

PΑ

SO

DT

LA IC

NCL

PΙ

ovarian tumor-assocd. markers are described for diagnosis, drug screening and therapy applications.

- ST ovary cancer marker gene profiling microarray comparative genomic hybridization; drug screening therapy diagnosis ovary cancer marker gene profiling
- IT Primers (nucleic acid) Probes (nucleic acid)

RL: ARG (Analytical reagent use); DGN (Diagnostic use); ANST (Analytical study); BIOL (Biological study); USES (Uses)

(5'-amino-linked degenerate oligonucleotide primer; methods for diagnosing and monitoring ovarian cancer by profiling assocd. marker genes using comparative genomic hybridization array)

IT Cyclins

RL: ANT (Analyte); BSU (Biological study, unclassified); DGN (Diagnostic use); PRP (Properties); ANST (Analytical study); BIOL (Biological study); USES (Uses)

(A, decreased expression in ovary cancer; methods for diagnosing and monitoring ovarian cancer by profiling assocd. marker genes using comparative genomic hybridization array)

IT Gene, animal

RL: ANT (Analyte); BSU (Biological study, unclassified); DGN (Diagnostic use); PRP (Properties); ANST (Analytical study); BIOL (Biological study); USES (Uses)

(ADRA1C; methods for diagnosing and monitoring ovarian cancer by profiling assocd. marker genes using comparative genomic hybridization array)

IT Gene, animal

RL: ANT (Analyte); BSU (Biological study, unclassified); DGN (Diagnostic use); PRP (Properties); ANST (Analytical study); BIOL (Biological study); USES (Uses)

(AF-4, decreased expression in ovary cancer; methods for diagnosing and monitoring ovarian cancer by profiling assocd. marker genes using comparative genomic hybridization array)

IT Gene, animal

RL: ANT (Analyte); BSU (Biological study, unclassified); DGN (Diagnostic use); PRP (Properties); ANST (Analytical study); BIOL (Biological study); USES (Uses)

(ATF4, decreased expression in ovary cancer; methods for diagnosing and monitoring ovarian cancer by profiling assocd. marker genes using comparative genomic hybridization array)

IT DNA microarray technology

(CGH (comparative genomic hybridization); methods for diagnosing and monitoring ovarian cancer by profiling assocd. marker genes using comparative genomic hybridization array)

IT Cadherins

RL: ANT (Analyte); BSU (Biological study, unclassified); DGN (Diagnostic use); PRP (Properties); ANST (Analytical study); BIOL (Biological study); USES (Uses)

(E-, decreased expression in ovary cancer; methods for diagnosing and monitoring ovarian cancer by profiling assocd. marker genes using comparative genomic hybridization array)

IT Gene, animal

RL: ANT (Analyte); BSU (Biological study, unclassified); DGN (Diagnostic use); PRP (Properties); ANST (Analytical study); BIOL (Biological study); USES (Uses)

(ERBB2, decreased expression in ovary cancer; methods for diagnosing and monitoring ovarian cancer by profiling assocd. marker genes using comparative genomic hybridization array)

IT Gene, animal

RL: ANT (Analyte); BSU (Biological study, unclassified); DGN (Diagnostic use); PRP (Properties); ANST (Analytical study); BIOL (Biological study); USES (Uses)

(Evi-1, increased expression in ovary cancer; methods for diagnosing

and monitoring ovarian cancer by profiling assocd. marker genes using comparative genomic hybridization array)

IT Gene, animal

RL: ANT (Analyte); BSU (Biological study, unclassified); DGN (Diagnostic use); PRP (Properties); ANST (Analytical study); BIOL (Biological study); USES (Uses)

(FGR, decreased expression in ovary cancer; methods for diagnosing and monitoring ovarian cancer by profiling assocd. marker genes using comparative genomic hybridization array)

IT Gene, animal

RL: ANT (Analyte); BSU (Biological study, unclassified); DGN (Diagnostic use); PRP (Properties); ANST (Analytical study); BIOL (Biological study); USES (Uses)

(FKHR, decreased expression in ovary cancer; methods for diagnosing and monitoring ovarian cancer by profiling assocd. marker genes using comparative genomic hybridization array)

IT Gene, animal

RL: ANT (Analyte); BSU (Biological study, unclassified); DGN (Diagnostic use); PRP (Properties); ANST (Analytical study); BIOL (Biological study); USES (Uses)

(GLUT2, increased expression in ovary cancer; methods for diagnosing and monitoring ovarian cancer by profiling assocd. marker genes using comparative genomic hybridization array)

IT Gene, animal

RL: ANT (Analyte); BSU (Biological study, unclassified); DGN (Diagnostic use); PRP (Properties); ANST (Analytical study); BIOL (Biological study); USES (Uses)

(HTR2A, decreased expression in ovary cancer; methods for diagnosing and monitoring ovarian cancer by profiling assocd. marker genes using comparative genomic hybridization array)

IT EST (expressed sequence tag)

RL: ANT (Analyte); BSU (Biological study, unclassified); DGN (Diagnostic use); PRP (Properties); ANST (Analytical study); BIOL (Biological study); USES (Uses)

(Hs.14518, related gene decreased expression in ovary cancer; methods for diagnosing and monitoring ovarian cancer by profiling assocd. marker genes using comparative genomic hybridization array)

IT Gene, animal

RL: ANT (Analyte); BSU (Biological study, unclassified); DGN (Diagnostic use); PRP (Properties); ANST (Analytical study); BIOL (Biological study); USES (Uses)

(INSR, decreased expression in ovary cancer; methods for diagnosing and monitoring ovarian cancer by profiling assocd. marker genes using comparative genomic hybridization array)

IT Gene, animal

RL: ANT (Analyte); BSU (Biological study, unclassified); DGN (Diagnostic use); PRP (Properties); ANST (Analytical study); BIOL (Biological study); USES (Uses)

(LPL, decreased expression in ovary cancer; methods for diagnosing and monitoring ovarian cancer by profiling assocd. marker genes using comparative genomic hybridization array)

IT Gene, animal

RL: ANT (Analyte); BSU (Biological study, unclassified); DGN (Diagnostic use); PRP (Properties); ANST (Analytical study); BIOL (Biological study); USES (Uses)

(RARA, decreased expression in ovary cancer; methods for diagnosing and monitoring ovarian cancer by profiling assocd. marker genes using comparative genomic hybridization array)

IT Gene, animal

RL: ANT (Analyte); BSU (Biological study, unclassified); DGN (Diagnostic use); PRP (Properties); ANST (Analytical study); BIOL (Biological study); USES (Uses)

(REV3L; methods for diagnosing and monitoring ovarian cancer by

profiling assocd. marker genes using comparative genomic hybridization array)

IT Gene, animal

RL: ANT (Analyte); BSU (Biological study, unclassified); DGN (Diagnostic use); PRP (Properties); ANST (Analytical study); BIOL (Biological study); USES (Uses)

(RGS, decreased expression in ovary cancer; methods for diagnosing and monitoring ovarian cancer by profiling assocd. marker genes using comparative genomic hybridization array)

IT Gene, animal

RL: ANT (Analyte); BSU (Biological study, unclassified); DGN (Diagnostic use); PRP (Properties); ANST (Analytical study); BIOL (Biological study); USES (Uses)

(SST, increased expression in ovary cancer; methods for diagnosing and monitoring ovarian cancer by profiling assocd. marker genes using comparative genomic hybridization array)

IT Transcription factors

RL: ANT (Analyte); BSU (Biological study, unclassified); DGN (Diagnostic use); PRP (Properties); ANST (Analytical study); BIOL (Biological study); USES (Uses)

(TBP (TATA box-binding protein), decreased expression in ovary cancer; methods for diagnosing and monitoring ovarian cancer by profiling assocd. marker genes using comparative genomic hybridization array)

IT Gene, animal

RL: ANT (Analyte); BSU (Biological study, unclassified); DGN (Diagnostic use); PRP (Properties); ANST (Analytical study); BIOL (Biological study); USES (Uses)

(TCL1 (T cell leukemia/lymphoma 1), decreased expression in ovary cancer; methods for diagnosing and monitoring ovarian cancer by profiling assocd. marker genes using comparative genomic hybridization array)

IT Gene, animal

RL: ANT (Analyte); BSU (Biological study, unclassified); DGN (Diagnostic use); PRP (Properties); ANST (Analytical study); BIOL (Biological study); USES (Uses)

(THRA, decreased expression in ovary cancer; methods for diagnosing and monitoring ovarian cancer by profiling assocd. marker genes using comparative genomic hybridization array)

IT Gene, animal

RL: ANT (Analyte); BSU (Biological study, unclassified); DGN (Diagnostic use); PRP (Properties); ANST (Analytical study); BIOL (Biological study); USES (Uses)

(TOP2A, decreased expression in ovary cancer; methods for diagnosing and monitoring ovarian cancer by profiling assocd. marker genes using comparative genomic hybridization array)

IT Gene, animal

RL: ANT (Analyte); BSU (Biological study, unclassified); DGN (Diagnostic use); PRP (Properties); ANST (Analytical study); BIOL (Biological study); USES (Uses)

(TP53, decreased expression in ovary cancer; methods for diagnosing and monitoring ovarian cancer by profiling assocd. marker genes using comparative genomic hybridization array)

IT Therapy

(adjuvant, for ovary cancer; methods for diagnosing and monitoring ovarian cancer by profiling assocd. marker genes using comparative genomic hybridization array)

IT Recombination, genetic

(amplification, of ovary cancer marker; methods for diagnosing and monitoring ovarian cancer by profiling assocd. marker genes using comparative genomic hybridization array)

IT mRNA

RL: ANT (Analyte); BSU (Biological study, unclassified); DGN (Diagnostic use); ANST (Analytical study); BIOL (Biological study); USES (Uses)

(assocd. with ovary cancer, detection of the level of; methods for diagnosing and monitoring ovarian cancer by profiling assocd. marker genes using comparative genomic hybridization array)

IT Diagnosis

(cancer; methods for diagnosing and monitoring ovarian cancer by profiling assocd. marker genes using comparative genomic hybridization array)

IT Mutation

(deletion, of ovary cancer marker; methods for diagnosing and monitoring ovarian cancer by profiling assocd. marker genes using comparative genomic hybridization array)

IT Neoplasm

(diagnosis; methods for diagnosing and monitoring ovarian cancer by profiling assocd. marker genes using comparative genomic hybridization array)

IT Animal tissue

Blood plasma

Blood serum

Cerebrospinal fluid

Saliva

(diagnostic sample; methods for diagnosing and monitoring ovarian cancer by profiling assocd. marker genes using comparative genomic hybridization array)

IT Antisense oligonucleotides

Ribozymes

RL: THU (Therapeutic use); BIOL (Biological study); USES (Uses) (for antitumor agent screening; methods for diagnosing and monitoring ovarian cancer by profiling assocd. marker genes using comparative genomic hybridization array)

IT Blood analysis

Urine analysis

(for diagnosis; methods for diagnosing and monitoring ovarian cancer by profiling assocd. marker genes using comparative genomic hybridization array)

IT Chemotherapy

Hormone replacement therapy

Immunotherapy

Radiotherapy

Surgery

(for ovary cancer; methods for diagnosing and monitoring ovarian cancer by profiling assocd. marker genes using comparative genomic hybridization array)

IT Gene, animal

RL: ANT (Analyte); BSU (Biological study, unclassified); DGN (Diagnostic use); PRP (Properties); ANST (Analytical study); BIOL (Biological study); USES (Uses)

(fyn, decreased expression in ovary cancer; methods for diagnosing and monitoring ovarian cancer by profiling assocd. marker genes using comparative genomic hybridization array)

IT Adrenoceptors

RL: ANT (Analyte); BSU (Biological study, unclassified); DGN (Diagnostic use); PRP (Properties); ANST (Analytical study); BIOL (Biological study); USES (Uses)

(gene ADRA1C, .alpha.1C, decreased expression in ovary cancer; methods for diagnosing and monitoring ovarian cancer by profiling assocd. marker genes using comparative genomic hybridization array)

IT Proteins

RL: ANT (Analyte); BSU (Biological study, unclassified); DGN (Diagnostic use); PRP (Properties); ANST (Analytical study); BIOL (Biological study); USES (Uses)

(gene ATF4, TAXREB67 protein, , decreased expression in ovary cancer; methods for diagnosing and monitoring ovarian cancer by profiling assocd. marker genes using comparative genomic hybridization array)

IT Proteins

RL: ANT (Analyte); BSU (Biological study, unclassified); DGN (Diagnostic use); PRP (Properties); ANST (Analytical study); BIOL (Biological study); USES (Uses)

(gene FKHR, forkhead protein; methods for diagnosing and monitoring ovarian cancer by profiling assocd. marker genes using comparative genomic hybridization array)

IT Platelet-derived growth factors

RL: ANT (Analyte); BSU (Biological study, unclassified); DGN (Diagnostic use); PRP (Properties); ANST (Analytical study); BIOL (Biological study); USES (Uses)

(gene PDGFB, decreased expression in ovary cancer; methods for diagnosing and monitoring ovarian cancer by profiling assocd. marker genes using comparative genomic hybridization array)

IT neu (receptor)

RL: ANT (Analyte); BSU (Biological study, unclassified); DGN (Diagnostic use); PRP (Properties); ANST (Analytical study); BIOL (Biological study); USES (Uses)

(gene c-erbB2, decreased expression in ovary cancer; methods for diagnosing and monitoring ovarian cancer by profiling assocd. marker genes using comparative genomic hybridization array)

IT Transcription factors

RL: ANT (Analyte); BSU (Biological study, unclassified); DGN (Diagnostic use); PRP (Properties); ANST (Analytical study); BIOL (Biological study); USES (Uses)

(gene myb, decreased expression in ovary cancer; methods for diagnosing and monitoring ovarian cancer by profiling assocd. marker genes using comparative genomic hybridization array)

IT Chromosome

(human 1, ovary cancer marker gene located at; methods for diagnosing and monitoring ovarian cancer by profiling assocd. marker genes using comparative genomic hybridization array)

IT Chromosome

(human 13, ovary cancer marker gene located at; methods for diagnosing and monitoring ovarian cancer by profiling assocd. marker genes using comparative genomic hybridization array)

IT Chromosome

(human 14, ovary cancer marker gene located at; methods for diagnosing and monitoring ovarian cancer by profiling assocd. marker genes using comparative genomic hybridization array)

IT Chromosome

(human 16, ovary cancer marker gene located at; methods for diagnosing and monitoring ovarian cancer by profiling assocd. marker genes using comparative genomic hybridization array)

IT Chromosome

(human 17, ovary cancer marker gene located at; methods for diagnosing and monitoring ovarian cancer by profiling assocd. marker genes using comparative genomic hybridization array)

IT Chromosome

(human 19, ovary cancer marker gene located at; methods for diagnosing and monitoring ovarian cancer by profiling assocd. marker genes using comparative genomic hybridization array)

IT Chromosome

(human 22, ovary cancer marker gene located at; methods for diagnosing and monitoring ovarian cancer by profiling assocd. marker genes using comparative genomic hybridization array)

IT Chromosome

(human 3, ovary cancer marker gene located at; methods for diagnosing and monitoring ovarian cancer by profiling assocd. marker genes using comparative genomic hybridization array)

IT Chromosome

(human 4, ovary cancer marker gene located at; methods for diagnosing and monitoring ovarian cancer by profiling assocd. marker genes using

comparative genomic hybridization array)

IT Chromosome

(human 6, ovary cancer marker gene located at; methods for diagnosing and monitoring ovarian cancer by profiling assocd. marker genes using comparative genomic hybridization array)

IT Chromosome

(human 8, ovary cancer marker gene located at; methods for diagnosing and monitoring ovarian cancer by profiling assocd. marker genes using comparative genomic hybridization array)

IT Antitumor agents

DNA sequences

Drug screening

Gene dosage

Nucleic acid hybridization

Ovary, neoplasm

Protein sequences

Susceptibility (genetic)

cDNA sequences

(methods for diagnosing and monitoring ovarian cancer by profiling assocd. marker genes using comparative genomic hybridization array)

IT Cheek

(mucosa, scraping, for diagnosis; methods for diagnosing and monitoring ovarian cancer by profiling assocd. marker genes using comparative genomic hybridization array)

IT Animal cell

(of ovary cancer, hyperproliferative; methods for diagnosing and monitoring ovarian cancer by profiling assocd. marker genes using comparative genomic hybridization array)

IT Genetic markers

Tumor markers

(of ovary cancer; methods for diagnosing and monitoring ovarian cancer by profiling assocd. marker genes using comparative genomic hybridization array)

IT Gene expression profiles, animal

(ovary cancer assocd.; methods for diagnosing and monitoring ovarian cancer by profiling assocd. marker genes using comparative genomic hybridization array)

IT Cat (Felis catus)

Cattle

Dog (Canis familiaris)

Horse (Equus caballus)

Human

Lagomorpha

Mouse

Nonhuman primate

Swine

(ovary cancer diagnosis in; methods for diagnosing and monitoring ovarian cancer by profiling assocd. marker genes using comparative genomic hybridization array)

IT Transcription factors

RL: THU (Therapeutic use); BIOL (Biological study); USES (Uses) (repressors, for antitumor agent screening; methods for diagnosing and monitoring ovarian cancer by profiling assocd. marker genes using comparative genomic hybridization array)

IT Cytotoxic agents

(screening for; methods for diagnosing and monitoring ovarian cancer by profiling assocd. marker genes using comparative genomic hybridization array)

IT Proteins

RL: ANT (Analyte); BSU (Biological study, unclassified); DGN (Diagnostic use); PRP (Properties); ANST (Analytical study); BIOL (Biological study); USES (Uses)

(solurshin, gene RGS, decreased expression in ovary cancer; methods for

diagnosing and monitoring ovarian cancer by profiling assocd. marker genes using comparative genomic hybridization array)

IT 5-HT receptors

RL: ANT (Analyte); BSU (Biological study, unclassified); DGN (Diagnostic use); PRP (Properties); ANST (Analytical study); BIOL (Biological study); USES (Uses)

(type 5-HT2A, gene HTR2A, decreased expression in ovary cancer; methods for diagnosing and monitoring ovarian cancer by profiling assocd. marker genes using comparative genomic hybridization array)

IT 115926-52-8, Phosphatidylinositol 3-kinase

RL: ANT (Analyte); BSU (Biological study, unclassified); DGN (Diagnostic use); PRP (Properties); ANST (Analytical study); BIOL (Biological study); USES (Uses)

(gene PIK3CA, increased expression in ovary cancer; methods for diagnosing and monitoring ovarian cancer by profiling assocd. marker genes using comparative genomic hybridization array)

IT 243664-63-3, DNA polymerase .zeta.

RL: ANT (Analyte); BSU (Biological study, unclassified); DGN (Diagnostic use); PRP (Properties); ANST (Analytical study); BIOL (Biological study); USES (Uses)

(gene REV3L, decreased expression in ovary cancer; methods for diagnosing and monitoring ovarian cancer by profiling assocd. marker genes using comparative genomic hybridization array)

TΤ 139805-78-0, GenBank M19722 140027-89-0, GenBank M15024 140035-35-4, 140035-45-6, GenBank J00306 144915-65-1, GenBank X57830 GenBank M12783 151066-51-2, GenBank D25235 164498-19-5, GenBank T79768 184512-11-6, GenBank U69961 252820-17-0, GenBank S82592 384442-63-1, GenBank X51688 384451-31-4, GenBank M10051 384463-70-1, GenBank M11730 384528-20-5, GenBank L13773 384597-53-9, GenBank X82240 389182-08-5, GenBank M14333 389182-26-7, GenBank Y00479 389185-22-2, GenBank M15856 391523-84-5, GenBank D90209 391530-10-2, GenBank J04088 391535-27-6, GenBank Z13009 391548-90-6, GenBank Z29090 392043-30-0, GenBank AF032885 398095-16-4, GenBank X06614 398425-77-9, GenBank M55654 412112-08-4, GenBank Z13009 RL: ANT (Analyte); BSU (Biological study, unclassified); DGN (Diagnostic use); PRP (Properties); ANST (Analytical study); BIOL (Biological study); USES (Uses)

(nucleotide sequence; methods for diagnosing and monitoring ovarian cancer by profiling assocd. marker genes using comparative genomic hybridization array)

IT 212216-75-6, GenBank AF078695

RL: BSU (Biological study, unclassified); PRP (Properties); BIOL (Biological study)

(nucleotide sequence; methods for diagnosing and monitoring ovarian cancer by profiling assocd. marker genes using comparative genomic hybridization array)

L117 ANSWER 6 OF 19 HCAPLUS COPYRIGHT 2003 ACS

AN 2002:555682 HCAPLUS

DN 137:104752

TI Probes to repeat sequence-free genomic regions for use in high throughput screening of genomes

IN Collins, Colin; Volik, Stanislav V.; Gray, Joe
W.; Albertson, Donna G.; Pinkel, Daniel

PA The Regents of the University of California, USA

SO PCT Int. Appl., 30 pp. CODEN: PIXXD2

DT Patent

LA English

IC ICM C12Q

CC 3-1 (Biochemical Genetics) Section cross-reference(s): 20

FAN.CNT 1

PATENT NO.

KIND DATE

APPLICATION NO. DATE

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PΙ
     WO 2002057481
                        A2
                              20020725
                                              WO 2002-US365
                                                                 20020107
          W: AE, AG, AL, AM, AT, AU, AZ, BA, BB, BG, BR, BY, BZ, CA, CH, CN,
              CO, CR, CU, CZ, DE, DK, DM, DZ, EC, EE, ES, FI, GB, GD, GE, GH,
              GM, HR, HU, ID, IL, IN, IS, JP, KE, KG, KP, KR, KZ, LC, LK, LR,
              LS, LT, LU, LV, MA, MD, MG, MK, MN, MW, MX, MZ, NO, NZ, OM, PH,
              PL, PT, RO, RU, SD, SE, SG, SI, SK, SL, TJ, TM, TN, TR, TT, TZ, UA, UG, UZ, VN, YU, ZA, ZM, ZW, AM, AZ, BY, KG, KZ, MD, RU, TJ,
         RW: GH, GM, KE, LS, MW, MZ, SD, SL, SZ, TZ, UG, ZM, ZW, AT, BE, CH, CY, DE, DK, ES, FI, FR, GB, GR, IE, IT, LU, MC, NL, PT, SE, TR, BF, BJ, CF, CG, CI, CM, GA, GN, GQ, GW, ML, MR, NE, SN, TD, TG
     US 2003022166
                         Α1
                              20030130
                                              US 2001-766450
                                                                 20010119
PRAI US 2001-766450
                        Α
                              20010119
     The present invention provides a rapid, efficient, and automated method
AB
     for identifying unique sequences within the genome. This invention
     involves the identification of repeat sequence-free subregions within a
     genomic region of interest as well as the detn. of which of those repeat
     sequence-free subregions are truly unique within the genome. Once the
     truly unique subregions are identified, primer sequences are generated
     that are suitable for the amplification of sequences, e.g., for use as
     probes or array targets, within the unique subregions.
ST
     oligonucleotide probe repeat sequence free human genome; primers non
     repeat sequence genome screening
IT
     Sequence homology analysis
         (BLAST, repeat sequences identified using; probes to repeat
        sequence-free genomic regions for use in high throughput screening of
IT
     DNA microarray technology
         (CGH, probes for; probes to repeat sequence-free genomic regions for
        use in high throughput screening of genomes)
ΙT
     Computer program
         (Repeat Masker or Primer 3 software, repeat sequences identified using;
        probes to repeat sequence-free genomic regions for use in high
        throughput screening of genomes)
IT
     Optical imaging devices
         (displaying oligonucleotide sequence on; probes to repeat sequence-free
        genomic regions for use in high throughput screening of genomes)
TΤ
     Human
         (genome; probes to repeat sequence-free genomic regions for use in high
        throughput screening of genomes)
IT
     Nucleic acid hybridization
        (in situ, fluorescence, non-repeat sequence probes for; probes to
        repeat sequence-free genomic regions for use in high throughput
        screening of genomes)
ΙT
        (nucleotide sequence; probes to repeat sequence-free genomic regions
        for use in high throughput screening of genomes)
IT
     Fluorescent substances
         (probe labeled with; probes to repeat sequence-free genomic regions for
        use in high throughput screening of genomes)
ΙT
     DNA sequences
     Genome
     Nucleic acid hybridization
         (probes to repeat sequence-free genomic regions for use in high
        throughput screening of genomes)
ΙT
     Probes (nucleic acid)
     RL: ARG (Analytical reagent use); BPN (Biosynthetic preparation); BUU
     (Biological use, unclassified); ANST (Analytical study); BIOL (Biological
     study); PREP (Preparation); USES (Uses)
        (probes to repeat sequence-free genomic regions for use in high
        throughput screening of genomes)
ΙT
     Repetitive DNA
     RL: BSU (Biological study, unclassified); BIOL (Biological study)
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(probes to repeat sequence-free genomic regions for use in high throughput screening of genomes)

ITPrimers (nucleic acid)

> RL: ARG (Analytical reagent use); BPN (Biosynthetic preparation); BUU (Biological use, unclassified); ANST (Analytical study); BIOL (Biological study); PREP (Preparation); USES (Uses)

(to non-repeat sequences of genome; probes to repeat sequence-free genomic regions for use in high throughput screening of genomes) IT443371-12-8 443371-13-9 443371-14-0 443371-15-1 443371-16-2 443371-17-3 443371-18-4 443371-19-5 443371-20-8 443371-21-9

443371-22-0 443371-23-1 443371-24-2 443371-26-4 443371-25-3 443371-27-5 443371-28-6 443371-29-7 443371-30-0 443371-31-1 443371-32-2 443371-33-3 443371-34-4 443371-35-5 443371-36-6 443371-37-7 443371-38-8 443371-39-9 443371-40-2 443371-41-3 443371-42-4 443371-43-5 443371-44-6 443371-45-7 443371-46-8 443371-47-9 443371-48-0 443371-49-1 443371-50-4 443371-51-5

443371-52-6 443371-53-7 443371-54-8 443371-55-9 443371-56-0 443371-57-1 443371-58-2 443371-59-3 443371-60-6 443371-61-7 443371-62-8 443371-63-9 443371-64-0 443371-65-1 443371-66-2 443371-67-3 443371-68-4 443371-69-5 443371-70-8 443371-71-9 443371-72-0 443371-73-1 443371-74-2 443371-75-3 443371-76-4

443371-77-5 443371-78-6 443371-79-7 443371-80-0 443371-81-1 443371-82-2 443371-83-3 443371-84-4 443371-85-5 443371-86-6 443371-87-7 443371-88-8 443371-89-9 443371-90-2 443371-91-3

443371-92-4 443371-93-5 443371-94-6 443371-95-7 443371-96-8 443371-97-9 443371-98-0 443371-99-1 443372-00-7 443372-01-8 443372-02-9 443372-03-0 443372-04-1 443372-05-2 443372-06-3

443372-07-4 443372-08-5 443372-09-6 443372-10-9 443372-11-0 443372-12-1 443372-13-2 443372-14-3 443372-15-4 443372-16-5 443372-19-8

443372-17-6 443372-18-7 443372-22-3 443372-23-4

RL: PRP (Properties)

(unclaimed sequence; probes to repeat sequence-free genomic regions for use in high throughput screening of genomes)

443372-20-1

443372-21**-**2

- L117 ANSWER 7 OF 19 HCAPLUS COPYRIGHT 2003 ACS
- 2001:895110 HCAPLUS
- DN 138:366457
- TΙ Genome scanning with array CGH delineates regional alterations in mouse islet. [Erratum to document cited in CA136:353395]
- ΑU Hodgson, Graeme.; Hager, Jeffrey H. H.; Volik, Stas.; Hariono, Sujatmi.; Wernick, Meredith.; Moore, Dan.; Nowak, Norma.; Albertson, Donna G.; Pinkel, Daniel; Collins, Colin; Hanahan, Douglas; Gray,
- CS Cancer Genetics and Breast Oncology Programs, UCSF Cancer Center, University of San Francisco at San Francisco, San Francisco, CA, 94143-0808, USA
- SO Nature Genetics (2001), 29(4), 491 CODEN: NGENEC; ISSN: 1061-4036
- PΒ Nature America Inc.
- DTJournal
- LAEnglish
- CC 14-1 (Mammalian Pathological Biochemistry) Section cross-reference(s): 3
- AΒ The name of the seventh author, Norma Nowak (Department of Human Genetics, Roswell Park Cancer Institute, Buffalo, new York 14263, USA), was omitted from the author list.
- ST erratum mouse pancreas tumor genome copy number; carcinoma mouse chromosome alteration LOH9 LOH16 erratum; microarray comparative genomic hybridization genome copy number erratum
- ΙT Nucleic acid hybridization

(DNA-DNA, CGH (comparative genomic hybridization); delineation of chromosomal alterations in mouse islet carcinomas using microarray-CGH, and identification of corresponding human genes (Erratum))

IT Gene, animal

RL: BSU (Biological study, unclassified); BIOL (Biological study) (FSTL1, PPP2R3, CDKNIB, STMN1, PFDN4 and CYP24; delineation of chromosomal alterations in mouse islet carcinomas using microarray-CGH, and identification of corresponding human genes (Erratum))

Pancreatic islet of Langerhans, neoplasm
(carcinoma; delineation of chromosomal alterations in mouse islet
carcinomas using microarray-CGH, and identification of corresponding
human genes (Erratum))

IT Gene dosage

(changes in; delineation of chromosomal alterations in mouse islet carcinomas using microarray-CGH, and identification of corresponding human genes (Erratum))

IT DNA microarray technology

Genome

Human

Loss of heterozygosity

Mouse

Mutation

(delineation of chromosomal alterations in mouse islet carcinomas using microarray-CGH, and identification of corresponding human genes (Erratum))

IT Chromosome

(mouse 14; delineation of chromosomal alterations in mouse islet carcinomas using microarray-CGH, and identification of corresponding human genes (Erratum))

IT Chromosome

(mouse 16, LOH16 region; delineation of chromosomal alterations in mouse islet carcinomas using microarray-CGH, and identification of corresponding human genes (Erratum))

IT Chromosome

(mouse 2; delineation of chromosomal alterations in mouse islet carcinomas using microarray-CGH, and identification of corresponding human genes (Erratum)) $\,$

IT Chromosome

(mouse 4; delineation of chromosomal alterations in mouse islet carcinomas using microarray-CGH, and identification of corresponding human genes (Erratum))

IT Chromosome

(mouse 6; delineation of chromosomal alterations in mouse islet carcinomas using microarray-CGH, and identification of corresponding human genes (Erratum))

IT Chromosome

(mouse 8; delineation of chromosomal alterations in mouse islet carcinomas using microarray-CGH, and identification of corresponding human genes (Erratum)) $\,$

IT Chromosome

(mouse 9, LOH9 region; delineation of chromosomal alterations in mouse islet carcinomas using microarray-CGH, and identification of corresponding human genes (Erratum))

L117 ANSWER 8 OF 19 HCAPLUS COPYRIGHT 2003 ACS

AN 2001:894920 HCAPLUS

DN 136:353395

TI Genome scanning with array CGH delineates regional alterations in mouse islet carcinomas

AU Hodgson, Graeme; Hager, Jeffrey H.; Volik, Stas; Hariono, Sujatmi; Wernick, Meredith; Moore, Dan; Albertson, Donna G.; Pinke, Daniel; Collins, Colin; Hanahan, Douglas; Gray, Joe W.

CS Cancer Genetics and Breast Onrology Programs, UCSF Cancer Center, University of California at San Francisco, San Francisco, CA, 94143-0808, USA

- SO Nature Genetics (2001), 29(4), 459-464 CODEN: NGENEC; ISSN: 1061-4036
- PB Nature America Inc.
- DT Journal
- LA English
- CC 14-1 (Mammalian Pathological Biochemistry) Section cross-reference(s): 3
- AΒ Carcinomas that develop in the pancreatic islets of transgenic mice expressing the SV40 T-antigens (Tag) under transcriptional control of the rat insulin II promoter (RIP) progress through well-characterized stages that are similar to aspects of human tumor progression, including hyperplastic growth, increased angiogenesis and reduced apoptosis. latter two stages have been assocd. with recurrent loss of heterozygosity (LOH) and reduced genome copy no. on chromosomes 9 (LOH9) and 16 (LOH16), aberrations which we believe contribute to these phenotypes. Earlier analyses localized LOH9 to approx. 3 Mb and LOH16 to approx. 30 Mb (both syntenic with human 3q21-q25) but were limited by low throughput and a lack of informative polymorphic markers. Here we show that comparative genomic hybridization to DNA microarrays (array CGH) overcomes these limitations by allowing efficient, genome-wide analyses of relative genome copy no. The CGH arrays used in these expts. carried BACs distributed at 2-20-MB intervals across the mouse genome and at higher d. in regions of interest. Using array CGH, we further narrowed the loci for LOH9 and LOH16 and defined new or previously unappreciated recurrent regions of copy-no. decrease on chromosomes 6, 8 and 14 (syntenic with human chromosomes 12p11-p13. 16q24.3 and 13q11-q32, resp.) and regions of copy-no. increase on chromosomes 2 and 4 (syntenic to human chromosomes 20q13.2 and 1p32-p36, resp.). Our analyses of human genome sequences syntenic to these regions suggest that CYP24, PFDN4, STMN1, CDKNIB, PPP2R3 and FSTL1 are candidate oncogenes or tumor-suppressor genes. We also show that irradn. and genetic background influence the spectrum of aberrations present in these tumors.
- ST mouse pancreas tumor genome copy number; carcinoma mouse chromosome alteration LOH9 LOH16; microarray comparative genomic hybridization genome copy number
- IT Nucleic acid hybridization

(DNA-DNA, CGH (comparative genomic hybridization); delineation of chromosomal alterations in mouse islet carcinomas using microarray-CGH, and identification of corresponding human genes)

IT Gene, animal

RL: BSU (Biological study, unclassified); BIOL (Biological study) (FSTL1, PPP2R3, CDKNIB, STMN1, PFDN4 and CYP24; delineation of chromosomal alterations in mouse islet carcinomas using microarray-CGH, and identification of corresponding human genes)

IT Pancreatic islet of Langerhans, neoplasm

(carcinoma; delineation of chromosomal alterations in mouse islet carcinomas using microarray-CGH, and identification of corresponding human genes)

IT Gene dosage

(changes in; delineation of chromosomal alterations in mouse islet carcinomas using microarray-CGH, and identification of corresponding human genes)

IT DNA microarray technology

Genome

Human

Loss of heterozygosity

Mouse

Mutation

(delineation of chromosomal alterations in mouse islet carcinomas using microarray-CGH, and identification of corresponding human genes)

IT Chromosome

(mouse 14; delineation of chromosomal alterations in mouse islet carcinomas using microarray-CGH, and identification of corresponding

human genes)

ΙT Chromosome

> (mouse 16, LOH16 region; delineation of chromosomal alterations in mouse islet carcinomas using microarray-CGH, and identification of corresponding human genes)

ΙΤ Chromosome

> (mouse 2; delineation of chromosomal alterations in mouse islet carcinomas using microarray-CGH, and identification of corresponding human genes)

IΤ Chromosome

> (mouse 4; delineation of chromosomal alterations in mouse islet carcinomas using microarray-CGH, and identification of corresponding

ΙT Chromosome

> (mouse 6; delineation of chromosomal alterations in mouse islet carcinomas using microarray-CGH, and identification of corresponding human genes)

IT Chromosome

> (mouse 8; delineation of chromosomal alterations in mouse islet carcinomas using microarray-CGH, and identification of corresponding human genes)

TΤ Chromosome

> (mouse 9, LOH9 region; delineation of chromosomal alterations in mouse islet carcinomas using microarray-CGH, and identification of corresponding human genes)

RE.CNT 25 THERE ARE 25 CITED REFERENCES AVAILABLE FOR THIS RECORD

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- (24) Vainberg, I; Cell 1998, V93, P863 HCAPLUS
- (25) Venter, J; Science 2001, V291, P1304 HCAPLUS
- L117 ANSWER 9 OF 19 HCAPLUS COPYRIGHT 2003 ACS
- ΑN 2001:886543 HCAPLUS
- DN 136:1600
- TΙ A novel method to identify rearrangements in a test genome from terminal sequence profiling
- ΤN Collins, Colin; Volik, Stanislav; Gray, Joe W.
- PΑ Regents of the University of California, USA
- SO PCT Int. Appl., 38 pp. CODEN: PIXXD2
- DTPatent

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English
IC
      ICM C120
CC
      3-1 (Biochemical Genetics)
      Section cross-reference(s): 14
FAN.CNT 1
      PATENT NO.
                        KIND
                              DATE
                                               APPLICATION NO. DATE
                        ____
                               _____
                                                -----
PΙ
     WO 2001092558
                         Α2
                               20011206
                                               WO 2001-US17757 20010531
     WO 2001092558
                         A3
                               20020404
              AE, AG, AL, AM, AT, AU, AZ, BA, BB, BG, BR, BY, BZ, CA, CH, CN,
              CO, CR, CU, CZ, DE, DK, DM, DZ, EC, EE, ES, FI, GB, GD, GE, GH, GM, HR, HU, ID, IL, IN, IS, JP, KE, KG, KP, KR, KZ, LC, LK, LR,
              LS, LT, LU, LV, MA, MD, MG, MK, MN, MW, MX, MZ, NO, NZ, PL, PT,
              RO, RU, SD, SE, SG, SI, SK, SL, TJ, TM, TR, TT, TZ, UA, UG, US, UZ, VN, YU, ZA, ZW, AM, AZ, BY, KG, KZ, MD, RU, TJ, TM
         RW: GH, GM, KE, LS, MW, MZ, SD, SL, SZ, TZ, UG, ZW, AT, BE, CH, CY, DE, DK, ES, FI, FR, GB, GR, IE, IT, LU, MC, NL, PT, SE, TR, BF, BJ, CF, CG, CI, CM, GA, GN, GW, ML, MR, NE, SN, TD, TG
     AU 2001075110
                         Α5
                               20011211
                                               AU 2001-75110
                                                                  20010531
PRAI US 2000-586529
                         Α
                               20000531
     WO 2001-US17757
                         W
                               20010531
AB
     The present invention provides a novel method to identify rearrangements
     in a test genome, e.g., a tumor genome, when compared to a ref. genome.
     This method provides major improvements over previous methods in terms of
     efficiency, rapidity, and cost-effectiveness. Briefly, this method
     involves generating or obtaining a large insert vector library from a test
     genome, sequencing the ends of the inserts in the library, and comparing
     the co-linearity of the sequenced ends in the library with corresponding
     sequences within a substantially-sequenced ref. genome. This invention is
     useful for any of a no. of applications, including for identifying
     rearrangements in tumor genomes and for detg. genetic differences between
     closely related species as well as between different strains of the same
     human tumor genome rearrangement terminal sequence profiling;
     bioinformatics computer software terminal sequence profiling
     Computer program
     Genetic vectors
     Genomic library
     PAC (P1-derived artificial chromosome)
         (a novel method to identify rearrangements in a test genome from
        terminal sequence profiling)
IΤ
     Insertion sequence
     RL: BSU (Biological study, unclassified); BIOL (Biological study)
         (a novel method to identify rearrangements in a test genome from
        terminal sequence profiling)
IT
     Neoplasm
        (cell, genome of, as test genome; a novel method to identify
        rearrangements in a test genome from terminal sequence profiling)
IT
     BAC (bacterial artificial chromosome)
        (clones; a novel method to identify rearrangements in a test genome
        from terminal sequence profiling)
ΙT
        (comparison; a novel method to identify rearrangements in a test genome
        from terminal sequence profiling)
IT
        (deletion; a novel method to identify rearrangements in a test genome
        from terminal sequence profiling)
IT
        (genome, as ref. genome; a novel method to identify rearrangements in a
        test genome from terminal sequence profiling)
IT
        (human, ref. genome present on different; a novel method to identify
```

rearrangements in a test genome from terminal sequence profiling)

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IT
     Bioinformatics
        (of terminal sequences; a novel method to identify rearrangements in a
        test genome from terminal sequence profiling)
IT
     Recombination, genetic
        (rearrangement, breakpoint; a novel method to identify rearrangements
        in a test genome from terminal sequence profiling)
ΙT
     Robotics
        (sequencing; a novel method to identify rearrangements in a test genome
        from terminal sequence profiling)
ΙT
     Genetic element
     RL: ANT (Analyte); BSU (Biological study, unclassified); ANST (Analytical
     study); BIOL (Biological study)
        (terminal sequence of clones; a novel method to identify rearrangements
        in a test genome from terminal sequence profiling)
ΙT
     Genetic methods
        (terminal sequence profiling; a novel method to identify rearrangements
        in a test genome from terminal sequence profiling)
ΙT
     DNA sequence analysis
        (terminal sequences; a novel method to identify rearrangements in a
        test genome from terminal sequence profiling)
IT
     Recombination, genetic
        (translocation; a novel method to identify rearrangements in a test
        genome from terminal sequence profiling)
L117 ANSWER 10 OF 19 HCAPLUS COPYRIGHT 2003 ACS
     2001:748135 HCAPLUS
AN
     135:268157
DN
TI
     DNA sequence analysis software
ΙN
     Collins, Colin; Volik, Stanislav; Gray, Joe W.
     The Regents of the University of California, USA
PA
     PCT Int. Appl., 40 pp.
SO
     CODEN: PIXXD2
DT
     Patent
LA
     English
     ICM G09G005-36
IC
     ICS C12Q001-68
CC
     3-1 (Biochemical Genetics)
     Section cross-reference(s): 20
FAN.CNT 1
     PATENT NO.
                      KIND DATE
                                           APPLICATION NO.
                                                            DATE
     WO 2001075856
                     A1
                            20011011
PΤ
                                           WO 2001-US10399 20010330
         W: AE, AG, AL, AM, AT, AU, AZ, BA, BB, BG, BR, BY, BZ, CA, CH, CN,
             CO, CR, CU, CZ, DE, DK, DM, DZ, EE, ES, FI, GB, GD, GE, GH, GM,
             HR, HU, ID, IL, IN, IS, JP, KE, KG, KP, KR, KZ, LC, LK, LR, LS,
             LT, LU, LV, MA, MD, MG, MK, MN, MW, MX, MZ, NO, NZ, PL, PT, RO,
             RU, SD, SE, SG, SI, SK, SL, TJ, TM, TR, TT, TZ, UA, UG, UZ, VN,
             YU, ZA, ZW, AM, AZ, BY, KG, KZ, MD, RU, TJ, TM
         RW: GH, GM, KE, LS, MW, MZ, SD, SL, SZ, TZ, UG, ZW, AT, BE, CH, CY,
             DE, DK, ES, FI, FR, GB, GR, IE, IT, LU, MC, NL, PT, SE, TR, BF,
             BJ, CF, CG, CI, CM, GA, GN, GW, ML, MR, NE, SN, TD, TG
                      . A
                            20000331
PRAI US 2000-541438
     A method and app, for analyzing a DNA sequence visually display important
     information about a sequence utilizes computer software. The invention
     discloses a software tool to identify and display repeat information
     (RepeatMasker), alignment information with sequences from public and
    proprietary data based, frequency of selected nucleotide pairs, and other
     information of interest to researchers.
ST
     computer software genetic method
IT
     Genetic element
     RL: ANT (Analyte); BSU (Biological study, unclassified); ANST (Analytical
     study); BIOL (Biological study)
```

(CpG island; DNA sequence anal. software)

```
ΙT
     Computer program
     Computers
     DNA sequence analysis
     DNA sequences
     Databases
     Génetic methods
     Mouse
     Repetitive DNA sequences
     Simulation and Modeling, biological
        (DNA sequence anal. software)
IT
     Genetic element
     Satellite DNA
     RL: ANT (Analyte); BSU (Biological study, unclassified); ANST (Analytical
     study); BIOL (Biological study)
        (DNA sequence anal. software)
ΙT
     Nucleic acid library
        (GSS; DNA sequence anal. software)
IT
     Repetitive DNA
     RL: ANT (Analyte); BSU (Biological study, unclassified); ANST (Analytical
     study); BIOL (Biological study)
        (LINE; DNA sequence anal. software)
TΤ
     Repetitive DNA
     RL: ANT (Analyte); BSU (Biological study, unclassified); ANST (Analytical
     study); BIOL (Biological study)
        (SINE; DNA sequence anal. software)
IT
     DNA sequences
        (alignment; DNA sequence anal. software)
IT
     Repetitive DNA
     RL: ANT (Analyte); BSU (Biological study, unclassified); ANST (Analytical
     study); BIOL (Biological study)
        (dinucleotide, CpG; DNA sequence anal. software)
TT
     Computer application
        (graphics; DNA sequence anal. software)
ΙT
     Genetic element
     RL: ANT (Analyte); BSU (Biological study, unclassified); ANST (Analytical
     study); BIOL (Biological study)
        (long terminal repeat; DNA sequence anal. software)
ΙT
     Genetic element
     RL: ANT (Analyte); BSU (Biological study, unclassified); ANST (Analytical
     study); BIOL (Biological study)
        (low complexity region; DNA sequence anal. software)
RE.CNT 2
              THERE ARE 2 CITED REFERENCES AVAILABLE FOR THIS RECORD
(1) McGall; US 6156501 A 2000 HCAPLUS
(2) Perlin; US 5876933 A 1999 HCAPLUS
L117 ANSWER 11 OF 19 HCAPLUS COPYRIGHT 2003 ACS
ΑN
     2001:429888 HCAPLUS
TΙ
     Comprehensive genome sequence analysis of a breast cancer amplicon
     Collins, Colin; Volik, Stanislav; Kowbel, David;
ΑU
     Ginzinger, David; Ylstra, Bauke; Cloutier, Thomas; Hawkins, Trevor;
     Predki, Paul; Martin, Christopher; Wernick, Meredith; Kuo, Wen-Lin;
     Alberts, Arthur; Gray, Joe W.
     University of California San Francisco Cancer Center, San Francisco, CA,
CS
     94143-0808, USA
SO
     Genome Research (2001), 11(6), 1034-1042
     CODEN: GEREFS; ISSN: 1088-9051
PB
     Cold Spring Harbor Laboratory Press
DT
     Journal; Letter
LA
     English
AΒ
     Gene amplification occurs in most solid tumors and is assocd. with poor
     prognosis. Amplification of 20q13.2 is common to several tumor types
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including breast cancer. The 1 Mb of sequence spanning the 20q13.2 breast

cancer amplicon is one of the most exhaustively studied segments of the human genome. These studies have included amplicon mapping by comparative genomic hybridization (CGH), fluorescent in-situ hybridization (FISH), array-CGH, quant. microsatellite anal. (QUMA), and functional genomic studies. Together these studies revealed a complex amplicon structure suggesting the presence of at least two driver genes in some tumors. One of these, ZNF217, is capable of immortalizing human mammary epithelial cells (HMEC) when overexpressed. In addn., we now report the sequencing of this region in human and mouse, and on quant. expression studies in tumors. Amplicon localization now is straightforward and the availability of human and mouse genomic sequence facilitates their functional anal. However, comprehensive annotation of megabase-scale regions requires integration of vast amts. of information. We present a system for integrative anal. and demonstrate its utility on 1.2 Mb of sequence spanning the 20q13.2 breast cancer amplicon and 865 kb of syntenic murine sequence. We integrate tumor genome copy no. measurements with exhausive genome landscape mapping, showing that amplicon boundaries are assocd. with maxima in repetitive element d. and a region of evolutionary instability. This integration of comprehensive sequence annotation, quant. expression anal., and tumor amplicon boundaries provide evidence for an addnl. driver gene prefoldin 4 (PFDN4), coregulated genes, conserved noncoding regions, and assoc. repetitive elements with regions of genomic instability at this locus.

RE CNT 32 THERE ARE 32 CITED REFERENCES AVAILABLE FOR THIS RECORD

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- (2) Beckman, M; Methods Enzymol 1997, V282, P200 HCAPLUS
- (3) Brosius, J; Gene 1999, V238, P115 HCAPLUS (4) Christian, S; Hum Mol Genet 1999, V8, P1025 HCAPLUS
- (5) Collins, C; Proc Natl Acad Sci 1998, V95, P8703 HCAPLUS
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- (7) Cuthill, S; Genes Chromosomes Cancer 1999, V26, P304 HCAPLUS
- (8) Eichler, E; Genome Res 1998, V8, P758 HCAPLUS
- (9) Eichler, E; Genome Res 1999, V9, P1048 HCAPLUS
- (10) Eichler, E; published erratum appears in Genome Res 1998, V10, P1095
- (11) Ginzinger, D; Cancer Res 2000, V60, P5405 HCAPLUS
- (12) Gray, J; Carcinogenesis 2000, V21, P443 HCAPLUS
- (13) Hansen, W; J Cell Biol 1999, V145, P265 HCAPLUS
- (14) Huie, M; Hum Genet 1999, V104, P94 HCAPLUS
- (15) Iijima, M; Acta Med Okayama 1996, V50, P73 HCAPLUS
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- (31) Vainberg, I; Cell 1998, V93, P863 HCAPLUS
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- L117 ANSWER 12 OF 19 HCAPLUS COPYRIGHT 2003 ACS
- 2000:790675 HCAPLUS ΑN
- DN 133:330496
- ΨT Comparative fluorescence hybridization to oligonucleotide microarrays used for precise mapping of chromosomal abnormalities assocd. with disease

```
Gray, Joe W.; Pinkel, Dan; Albertson, Donna G.; Collins,
     Colin C.; Baldocchi, Russell A.
PΑ
     The Regents of the University of California, USA
SO
     PCT Int. Appl., 28 pp.
     CODEN: PIXXD2
DT
     Patent
LA
     English
IC
     ICM C12Q001-68
     ICS C12P019-34; C07H021-02; C07H021-04
     3-1 (Biochemical Genetics)
     Section cross-reference(s): 14
FAN.CNT 1
     PATENT NO.
                      KIND
                                           APPLICATION NO.
                                                             DATE
     WO 2000066779
                            20001109
PΤ
                       Α1
                                           WO 2000-US11433
                                                            20000428
         W: CA, JP
         RW: AT, BE, CH, CY, DE, DK, ES, FI, FR, GB, GR, IE, IT, LU, MC, NL,
             PT, SE
     US 6465182
                            20021015
                       В1
                                           US 1999-302056
                                                             19990429
     EP 1173621
                       Α1
                            20020123
                                           EP 2000-930197
                                                             20000428
         R: AT, BE, CH, DE, DK, ES, FR, GB, GR, IT, LI, LU, NL, SE, MC, PT,
             IE, FI
PRAI US 1999-302056
                            19990429
                       Α
     WO 2000-US11433
                            20000428
                       W
AΒ
     The present invention provides methods of detg. relative copy no. of
     target nucleic acid sequences and precise mapping of chromosomal
     abnormalities assocd. with disease. The methods of the invention use
     target nucleic acid sequences immobilized on a solid surface, to which a
     sample comprising two sets of differentially labeled nucleic acid
     sequences are hybridized. The hybridization of the labeled nucleic acid
     sequences to the solid surface is then detected using std. techniques.
     nucleic acid array comparative fluorescence hybridization oligonucleotide
     probe; disease diagnosis chromosome aberration mapping DNA microarray
     fluorescence hybridization; tumor diagnosis DNA comparative fluorescence
     hybridization
ΙΤ
     RL: ARG (Analytical reagent use); BUU (Biological use, unclassified); THU
     (Therapeutic use); ANST (Analytical study); BIOL (Biological study); USES
        (COT-1, blocking agent; comparative fluorescence hybridization to
        oligonucleotide microarrays used for precise mapping of chromosomal
        abnormalities assocd. with disease)
IT
     Chromosome aberrations
     Diagnosis
     Genetic mapping
     Immobilization, biochemical
     Indicators
     Nucleic acid hybridization
        (comparative fluorescence hybridization to oligonucleotide microarrays
        used for precise mapping of chromosomal abnormalities assocd. with
        disease)
TΤ
     Nucleic acids
     RL: ANT (Analyte); BUU (Biological use, unclassified); THU (Therapeutic
     use); ANST (Analytical study); BIOL (Biological study); USES (Uses)
        (comparative fluorescence hybridization to oligonucleotide microarrays
        used for precise mapping of chromosomal abnormalities assocd. with
        disease)
    Gene, animal
ΙT
     RL: ANT (Analyte); THU (Therapeutic use); ANST (Analytical study); BIOL
     (Biological study); USES (Uses)
```

(comparative fluorescence hybridization to oligonucleotide microarrays used for precise mapping of chromosomal abnormalities assocd. with

disease)

IT cDNA

RL: ARG (Analytical reagent use); BUU (Biological use, unclassified); THU (Therapeutic use); ANST (Analytical study); BIOL (Biological study); USES (Uses)

(comparative fluorescence hybridization to oligonucleotide microarrays used for precise mapping of chromosomal abnormalities assocd. with disease)

IT mRNA

RL: BUU (Biological use, unclassified); BIOL (Biological study); USES (Uses)

(comparative fluorescence hybridization to oligonucleotide microarrays used for precise mapping of chromosomal abnormalities assocd. with disease)

IT Animal tissue

(fetal; comparative fluorescence hybridization to oligonucleotide microarrays used for precise mapping of chromosomal abnormalities assocd. with disease)

IT Embryo, animal

(fetus, tissue of; comparative fluorescence hybridization to oligonucleotide microarrays used for precise mapping of chromosomal abnormalities assocd. with disease)

IT Disease, animal

(genetic; comparative fluorescence hybridization to oligonucleotide microarrays used for precise mapping of chromosomal abnormalities assocd. with disease)

IT Glass beads

RL: NUU (Other use, unclassified); THU (Therapeutic use); BIOL (Biological study); USES (Uses)

(immobilization surface; comparative fluorescence hybridization to oligonucleotide microarrays used for precise mapping of chromosomal abnormalities assocd. with disease)

IT Nucleic acid hybridization

(in situ, fluorescence; comparative fluorescence hybridization to oligonucleotide microarrays used for precise mapping of chromosomal abnormalities assocd. with disease)

IT Probes (nucleic acid)

RL: ARG (Analytical reagent use); BUU (Biological use, unclassified); THU (Therapeutic use); ANST (Analytical study); BIOL (Biological study); USES (Uses)

(labeled, with fluorescent indicator; comparative fluorescence hybridization to oligonucleotide microarrays used for precise mapping of chromosomal abnormalities assocd. with disease)

IT Neoplasm

(nucleic acid from; comparative fluorescence hybridization to oligonucleotide microarrays used for precise mapping of chromosomal abnormalities assocd. with disease)

IT PCR (polymerase chain reaction)

(used to prep. labeled nucleic acid sequences; comparative fluorescence hybridization to oligonucleotide microarrays used for precise mapping of chromosomal abnormalities assocd. with disease)

RE.CNT 3 THERE ARE 3 CITED REFERENCES AVAILABLE FOR THIS RECORD

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- (2) Lisitsyn; Science 1993, V259, P946 HCAPLUS
- (3) Pinkel; US 5830645 A 1998 HCAPLUS

L117 ANSWER 13 OF 19 HCAPLUS COPYRIGHT 2003 ACS

AN 2000:397353 HCAPLUS

DN 133:291649

- TI Quantitative mapping of amplicon structure by array CGH identifies CYP24 as a candidate oncogene
- AU Albertson, Donna G.; Ylstra, Bauke; Segraves, Richard; Collins,

```
Colin; Dairkee, Shanaz H.; Kowbel, David; Kuo, Wen-Lin; Gray,
     Joe W.; Pinkel, Daniel
     Cancer Research Institute, University of California, San Francisco, San
     Francisco, CA, USA
     Nature Genetics (2000), 25(2), 144-146
SO
     CODEN: NGENEC; ISSN: 1061-4036
     Nature America Inc.
PB
DT
     Journal
LA
     English
CC
     3-1 (Biochemical Genetics)
     Section cross-reference(s): 14
     The authors show here that the quant, measurement of DNA copy no. across
AΒ
     amplified regions using array comparative genomic hybridization GCH may
     facilitate oncogene identification by providing precise information on the
     locations of both amplicon boundaries and amplification maxima.
ST
     oncogene map amplicon
ΙT
     Gene, animal
     RL: BOC (Biological occurrence); BSU (Biological study, unclassified);
     BIOL (Biological study); OCCU (Occurrence)
        (oncogene; quant. mapping of amplicon structure by array CGH identifies
        CYP24 as candidate oncogene)
TΤ
     Genetic mapping
        (quant. mapping of amplicon structure by array CGH identifies CYP24 as
        candidate oncogene)
ΙT
     RL: BOC (Biological occurrence); BSU (Biological study, unclassified);
     BIOL (Biological study); OCCU (Occurrence)
        (quant. mapping of amplicon structure by array CGH identifies CYP24 as
        candidate oncogene)
RE.CNT
        15
              THERE ARE 15 CITED REFERENCES AVAILABLE FOR THIS RECORD
(1) Albertson, D; Caenorhabditis elegans: Modern Biological Analysis of an
    Organism 1995, P339 HCAPLUS
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L117 ANSWER 14 OF 19 HCAPLUS COPYRIGHT 2003 ACS
ΑN
     2000:203128 HCAPLUS
DN
     132:330389
     Genome changes and gene expression in human solid tumors
ΤI
AU
     Gray, Joe W.; Collins, Colin
     UCSF Cancer Center, University of California San Francisco, San Francisco,
CS
     CA, 94143-0808, USA
SO
     Carcinogenesis (2000), 21(3), 443-452
     CODEN: CRNGDP; ISSN: 0143-3334
PB
     Oxford University Press
DT
     Journal; General Review
LA
     English
CC
     3-0 (Biochemical Genetics)
```

A review, with 141 refs. Genome-wide anal. techniques such as chromosome

Section cross-reference(s): 14

AΒ

painting, comparative genomic hybridization, representational difference anal., restriction landmark genome scanning and high-throughput anal. of LOH are now accelerating high-resoln. genome aberration localization in human tumors. These techniques are complemented by procedures for detection of differentially expressed genes such as differential display, nucleic acid subtraction, serial anal. of gene expression and expression microarray anal. These efforts are enabled by work from the human genome program in phys. map development, cDNA library prodn./sequencing, and genome sequencing. This review covers several commonly used large-scale genome and gene expression anal. techniques, outlines genomic approaches to gene discovery and summarizes information that has come from large-scale analyses of human solid tumors.

ST review genetic technique tumor

IT Genetic methods Genome

(genome changes and gene expression in human solid tumors)

IT Gene, animal

RL: ADV (Adverse effect, including toxicity); BPR (Biological process); BSU (Biological study, unclassified); BIOL (Biological study); PROC (Process)

(genome changes and gene expression in human solid tumors)

IT Chromosome

(human; genome changes and gene expression in human solid tumors)
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- L117 ANSWER 15 OF 19 HCAPLUS COPYRIGHT 2003 ACS
- AN 1998:798281 HCAPLUS
- DN 130:192338
- TI Genome scanning and gene discovery in breast and ovarian cancer
- ΑU Gray, Joe W.; Collins, Colin; Pinkel, Daniel; Shayesteh, Laleh; Lu, Yiling; Mills, Gordon
- UCSF Cancer Center, University of California, San Francisco, CA, USA CS
- SO Pezcoller Foundation Symposia (1998), 9(Biology of Tumors), 65-72 CODEN: PFSYES; ISSN: 0961-785X
- PΒ Plenum Publishing Corp.
- DTJournal; General Review
- LAEnglish
- CC 3-0 (Biochemical Genetics) Section cross-reference(s): 14
- AΒ A review, with 26 refs., on the use of comparative genomic hybridization (CGH) to scan breast and ovarian cancers. Recurrent abnormalities were defined in these studies. The authors focused on (1) a region of increased copy no. on chromosome 20q esp. apparent in breast cancers, and (2) a region of increased copy no. at chromosome 3q26 esp. apparent in ovarian cancers.

borin - 09 / 586529 STreview genome scanning breast ovary cancer ITGene dosage Genome Ovary, neoplasm (genome scanning and gene discovery in human breast and ovarian cancer) ΙT Chromosome (human 20; genome scanning and gene discovery in human breast and ovarian cancer) ΙT Chromosome (human 3; genome scanning and gene discovery in human breast and ovarian cancer) ΙT Genetics (hybridization, comparative genomic hybridization; genome scanning and gene discovery in human breast and ovarian cancer) IT Mammary gland (neoplasm; genome scanning and gene discovery in human breast and ovarian cancer) RE.CNT THERE ARE 26 CITED REFERENCES AVAILABLE FOR THIS RECORD (1) Bockmuhl, U; Laryngorhinootologie 1996, V75, P408 MEDLINE (2) Chang, H; Science 1997, V276, P1848 HCAPLUS (3) Cheng, J; Proc Natl Acad Sci USA 1992, V89, P9267 HCAPLUS (4) Church, D; Nat Genet 1994, V6, P98 HCAPLUS (5) Collins, C; Proceedings of the National Academy of Science (USA) (submitted) 1998 (6) Dietrich, W; Proceedings of the National Academy of Science (USA) 1994, V91, P9451 HCAPLUS (7) Heselmyer, K; Proc Natl Acad Sci USA 1996, V93, P479 (8) Isola, J; Am J Pathol 1995, V147, P905 MEDLINE (9) Iwabuchi, H; Cancer Research 1995, V55, P6172 HCAPLUS (10) Kallioniemi, A; Porch Natl Acad Sci U S A 1994, V91, P2156 HCAPLUS (11) Kallioniemi, A; Science 1992, V258, P818 HCAPLUS (12) Kimmerly, W; Genet Anal Tech Appl 1994, V11, P117 HCAPLUS (13) Levin, N; Genes Chromosomes and Cancer 1995, V13, P175 HCAPLUS (14) Lovett, M; Proc Natl Acad Sci U S A 1991, V88, P9628 HCAPLUS (15) Marte, B; Trends Biochem Sci 1997, V22, P355 HCAPLUS (16) Mohapatra, G; Genes Chromosomes Cancer 1995, V13, P86 HCAPLUS (17) Pinkel, D; Nature Genetics (submitted) 1998 (18) Reznikoff, C; Genes Dev 1994, V8, P2227 HCAPLUS (19) Savelieva, E; Oncogene 1997, V14, P551 HCAPLUS (20) Schlegel, J; Cancer Res 1995, V55, P6002 HCAPLUS (21) Shayesteh, L; Nature Genetics (submitted) 1998 (22) Solinas-Toldo, S; Cancer Res 1996, V56, P3803 HCAPLUS (23) Solinas-Toldo, S; Proc Natl Acad Sci U S A 1997, V94, P3854 HCAPLUS (24) Speicher, M; Cancer Research 1995, V55, P1010 HCAPLUS (25) Tanner, M; Cancer Res 1994, V54, P4257 HCAPLUS (26) Tanner, M; Clinical Cancer Research 1995, V1, P1455 HCAPLUS L117 ANSWER 16 OF 19 HCAPLUS COPYRIGHT 2003 ACS

AN 1998:663115 HCAPLUS

DN 130:21094

- High resolution analysis of DNA copy number variation using comparative genomic hybridization to microarrays
- ΑU Pinkel, Daniel; Segraves, Richard; Sudar, Damir; Clark, Steven; Poole, Ian; Kowbel, David; Collins, Colin; Kuo, Wen-Lin; Chen, Chira; Zhai, Ye; Dairkee, Shanaz H.; Ljung, Britt-marie; Gray, Joe W.; Albertson, Donna G.
- CS Cancer Genetics Program, UCSF Cancer Center, Univ. California, San Francisco, CA, 94143-0808, USA
- SO Nature Genetics (1998), 20(2), 207-211 CODEN: NGENEC; ISSN: 1061-4036
- Nature America PΒ
- DT Journal

- LA English
- CC 3-1 (Biochemical Genetics)
 Section cross-reference(s): 14
- Gene dosage variations occur in many diseases. In cancer, deletions and copy no. increases contribute to alterations in the expression of tumor-suppressor genes and oncogenes, resp. Developmental abnormalities, such as Down, Prader Willi, Angelman and Cri du Chat syndromes, result from gain or loss of one copy of a chromosome or chromosomal region. Thus, detection and mapping of copy no. abnormalities provide and approach for assocg. aberrations with disease phenotype and for localizing crit. genes. Comparative genomic hybridization (CGH) was developed for genome-wide anal. of DNA sequence copy no. in a single expt. In CGH, differentially labeled total genomic DNA from a 'test' and a 'ref.' cell population are co-hybridized to normal metaphase chromosomes, using blocking DNA to suppress signals from repetitive sequences. The resulting ratio of the fluorescence intensities at a location on the 'cytogenetic map', provided by the chromosomes, is approx. proportional to the ratio of the copy nos. of the corresponding DNA sequences in the test and ref. genomes. CGH has been broadly applied to human and mouse malignancies. The use of metaphase chromosomes, however, limits detection of events involving small regions (of less than 20 Mb) of the genome, resoln. of closely spaced aberrations and linking ratio changes to genomic/genetic markers. Therefore, more laborious locus-by-locus techniques have been required for higher resoln. studies. Hybridization to an array of mapped sequences instead of metaphase chromosomes could overcome the limitations of conventional CGH (ref. 6) if adequate performance could be achieved. Copy no. would be related to the test/ref. fluorescence ratio on the array targets, and genomic resoln. could be detd. by the map distance between the targets, or by the length of the cloned DNA segments. We describe here our implementation of array CGH. We demonstrate its ability to measure copy no. with high precision in the human genome, and to analyze clin. specimens by obtaining new information on chromosome 20 aberrations in breast cancer.
- ST comparative genomic hybridization chromosome aberration human breast cancer
- IT Nucleic acid hybridization

(DNA-DNA; high resoln. anal. of DNA copy no. variation using comparative genomic hybridization to microarrays)

IT Genetic methods

(comparative genomic hybridization; high resoln. anal. of DNA copy no. variation using comparative genomic hybridization to microarrays)

IT Chromosome aberrations

Genetic mapping

(high resoln. anal. of DNA copy no. variation using comparative genomic hybridization to microarrays)

IT Chromosome

(human 20; high resoln. anal. of DNA copy no. variation using comparative genomic hybridization to microarrays)

IT Mammary gland

(neoplasm; high resoln. anal. of DNA copy no. variation using comparative genomic hybridization to microarrays)

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- (31) Wittrup, K; Cytometry 1994, V16, P206 MEDLINE
- L117 ANSWER 17 OF 19 HCAPLUS COPYRIGHT 2003 ACS
- 1992:208828 HCAPLUS ΑN
- DN 116:208828
- ΤI Construction and characterization of plasmid libraries enriched in sequences from single human chromosomes
- ΑU Collins, Colin; Kuo, Wen Lin; Segraves, Richard; Fuscoe, James; Pinkel, Daniel; Gray, Joe W.
- CS Biomed. Sci. Div., Lawrence Livermore Natl. Lab., Livermore, CA, 94550,
- SO Genomics (1991), 11(4), 997-1006 CODEN: GNMCEP; ISSN: 0888-7543
- DΨ Journal
- LAEnglish
- CC3-2 (Biochemical Genetics) Section cross-reference(s): 13
- Plasmid libraries enriched in sequences from single chromosome types have AR been constructed for all human chromosomes. This was accomplished by transferring inserts from the Charon 21A phage libraries constructed by the National Lab. Gene Library Project into Bluescribe plasmids. Insert material freed by complete digestion of the phage libraries with HindIII or EcoRI was cloned into the corresponding sites in Bluescribe plasmids. The sizes of the Bluescribe library inserts detd. by gel electrophoresis range from near 0 to .apprx.6 kb. Fluorescence in situ hybridization (FISH) with the plasmid libraries showed that all hybridize along both arms of the expected (target) chromosome type with varying intensity. However, the plasmid libraries for chromosomes 1, 4, 9, 11, 16, 18, and 20 hybridize weakly or not at all near the centromeres of the target chromosome types. The libraries for chromosomes 13, 14, 15, 21, and 22 cross-hybridize near the centromeres of all members of this group and hybridize weakly to the short arms of the target chromosomes. FISH with each library allows specific staining of the target chromosome type in metaphase spreads. The signals resulting from FISH with libraries for chromosome 1, 4, 8, 9, 13, 14, 17, 18, 21, and Y are sufficiently intense to permit anal. in interphase nuclei. Examples of the use of these
- and interphase aneuploidy anal. are presented. human chromosome cloning Bluescribe plasmid; fluorescence in situ ST hybridization human chromosome; plasmid library single human chromosome ΙT Plasmid and Episome
 - (Bluescribe, contg. enriched sequences from single human chromosomes,

libraries for translocation detection, marker chromosome characterization,

construction and characterization of)

IT Interphase, biological

(aneuploidy of, anal. of human, using Bluescribe plasmid libraries enriched in sequences from single chromosome types)

IT Chromosome

(cloning of enriched sequences from single human, in Bluescribe plasmids, anal. of chromosome aberrations subsequent to)

IT Molecular cloning

(of enriched sequences from single human chromosomes, in Bluescribe vectors, chromosome aberration anal. subsequent to)

IT Nucleic acid hybridization

(in situ, fluorescence, in detection of human chromosome aberrations, using Bluescribe plasmid libraries enriched in sequences from single chromosome types)

IT Recombination, genetic

(translocation, detection of human chromosome, using Bluescribe plasmid libraries enriched in sequences from single chromosome types)

L117 ANSWER 18 OF 19 HCAPLUS COPYRIGHT 2003 ACS

AN 1989:547970 HCAPLUS

DN **111:147970**

- TI An efficient method for selecting unique-sequence clones from DNA libraries and its application to fluorescent staining of human chromosome 21 using in situ hybridization
- AU Fuscoe, James C.; Collins, Colin C.; Pinkel, Dan; Gray, Joe W.
- CS Biomed. Sci. Div., Lawrence Livermore Natl. Lab., Livermore, CA, 94550,
- SO Genomics (1989), 5(1), 100-9 CODEN: GNMCEP; ISSN: 0888-7543

DT Journal

LA English

CC 3-5 (Biochemical Genetics)
 Section cross-reference(s): 13

- An efficient procedure is described for selecting large nos. of AΒ unique-sequence or very low repeat-sequence probes from recombinant phage libraries. Probes were selected from the Charon 21A library LL21NSO2 (made from DNA from human chromosome 21) in a multistep process is in which (1) inserts from LL21NSO2 were subcloned into Blue-scribe plasmids, (2) plasmids were grown at high d. in colonies on nitrocellulose, and (3) plasmids were selected as contg. unique-sequence inserts if DNA from the colonies failed to hybridize, at low stringency, to radiolabeled total human DNA. Thus, 1530 colonies were picked to form the library pBS-U21/1530. About 80% of the recombinants constituting pBS-U21/1530 contained inserts that are present in only one copy in haploid genomic human DNA. Approx. 70% of the sequences mapped to human chromosome 21. Fluorescence in situ hybridization with DNA from pBS-U21/1530 allowed specific, intense staining of the no. 21 chromosomes in metaphase spreads made from human lymphocytes.
- ST DNA unique sequence clone selection; sequence DNA low copy repeat clone; human chromosome 21 mapping unique sequence

IT Deoxyribonucleic acid sequences

(unique, method for selecting cloned, from DNA libraries, fluorescence in situ hybridization of human chromosome 21 in relation to)

IT Chromosome

(human 21, unique-sequence DNA on, mapping of, fluorescence in situ hybridization for)

IT Deoxyribonucleic acids

RL: BIOL (Biological study)

(repetitive, low copy no., method for selecting cloned, from DNA libraries, human chromosome 21 in relation to)

```
AN
     1989:89879 HCAPLUS
DN
     110:89879
TI
     Fluorescence in situ hybridization with human chromosome-specific
     libraries: detection of trisomy 21 and translocations of chromosome 4
ΑU
     Pinkel, D.; Landegent, J.; Collins, C.; Fuscoe, J.; Segraves,
     R.; Lucas, J.; Gray, J.
CS
     Biomed. Sci. Div., Lawrence Livermore Natl. Lab., Livermore, CA, 94550,
SO
     Proceedings of the National Academy of Sciences of the United States of
     America (1988), 85(23), 9138-42
     CODEN: PNASA6; ISSN: 0027-8424
DT
     Journal
LA
     English
CC
     3-5 (Biochemical Genetics)
     Section cross-reference(s): 13, 14
     Chromosomes can be specifically stained in metaphase spreads and
AΒ
     interphase nuclei by in situ hybridization with entire chromosome-specific
     DNA libraries. Unlabeled human genomic DNA is used to inhibit the
     hybridization of sequences in the library that bind to multiple
     chromosomes. The target chromosome can be made at least 20 times brighter
     per unit length than the others. Trisomy 21 and translocations involving
     chromosome 4 can be detected in metaphase spreads and interphase nuclei by
     using this technique.
ST
     human chromosome detection fluorescence hybridization; trisomy 21
     detection human fluorescence hybridization; chromosome 4 translocation
     detection fluorescence hybridization
IT
     Chromosome
        (human 21, trisomy, detection of, by fluorescence in situ
        hybridization)
IT
     Nucleic acid hybridization
        (in situ, fluorescence, for human trisomy 21 and chromosome 4
        translocation detection)
ΙT
     Recombination, genetic
        (translocation, chromosome 4, of human, fluorescence in situ
        hybridization for detection of)
=> d his
     (FILE 'HOME' ENTERED AT 15:19:11 ON 15 JUL 2003)
                SET COST OFF
     FILE 'MEDLINE' ENTERED AT 15:19:49 ON 15 JUL 2003
                E COLLINS C/AU
            807 S E3-E23, E51-E53
L1
                E VOLIK S/AU
L_2
             12 S E3-E5
                E GRAY J/AU
L3
            689 S E3, E30, E31
                E GRAY JOE/AU
L4
             10 S E3, E4
L5
             15 S END SEQUENC? (L) PROFIL?
L6
            966 S ESP
L7
              1 S L1-L4 AND L5, L6
                E SEQUENCE ANALYSIS/CT
                E E3+ALL
\Gamma8
         878518 S E3+NT
```

L9

L10

L11

L12

L13

T₁14

734719 S L8 AND PY<=2000

482 S END SEQUENC?

265 S L1-L4 AND L8

235 S L1-L4 AND L9

292534 S L10, L11 AND PY<=2000

342230 S TERMIN?

```
L15
              1 S L13, L14 AND L10
L16
              25 S L13, L14 AND L11
                 E COMPUTATION/CT
                 E E9+ALL
L17
            3603 S E6+NT
L18
             531 S L17 AND L9
L19
              46 S L17 AND L12
                 SEL DN AN 6
               1 S E1-E3
L20
               3 S L7, L15, L20
L21
                 E GENOME/CT
                 E E3+ALL
                 E E5+ALL
L22
          97074 S E4+NT AND L9
                 E CHROMOSOM/CT
                E E6+ALL
                E E2+ALL
L23
          41888 S E8+NT AND L9
                 E E59+ALL
L24
           1883 S L9 AND E54+NT
           6094 S L9 AND E55+NT
L25
                E CLONE/CT
                E E25+ALL
                E E2+ALL
L26
          35529 S E4+NT
                E CLONING, MOLECULAR/CT
                E E3+ALL
L27
         117398 S E4+NT
                E E10+ALL
L28
           2025 S E9+NT
                E E24+ALL
                E E11+ALL
L29
          45092 S E5+NT
                E E23+ALL
          32772 S E17+NT
L30
                E E40+ALL
                E E22+ALL
L31
          71911 S E4+NT
                E E28+ALL
L32
          47762 S E5+NT
L33
          31993 S E16+NT OR E17+NT
L34
         170652 S L9 AND L26-L33
L35
         150092 S L9 AND CLON?
         206915 S L34, L35
L36
L37
           7716 S L23 AND L36
                E SEQUENCE ANALYSIS+ALL/CT
L38
          47793 S E4+NT AND PY<=2000
L39
           3546 S L38 NOT AB/FA
L40
          44247 S L38 NOT L39
L41
             55 S L40 AND L10
L42
           9854 S L40 AND L11
                SEL DN AN L41 7 16 28 38 41 44
L43
              6 S L41 AND E1-E18
L44
              9 S L21, L43 AND L1-L43
L45
              9 S L42 AND L17
L46
           2105 S L42 AND L22
L47
             95 S L42 AND L23
L48
             36 S L42 AND L24, L25
L49
           3542 S L42 AND L26-L33
L50
           3648 S L42 AND CLON?
L51
           5342 S L45-L50
L52
            95 S L23 AND L51
             13 S L52 AND BREAKPOINT
L53
```

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E CHROMSOME BREAKAGE/CT
                 E CHROMOSOME BREAKAGE/CT
                 E E3+LL
                E E3+ALL
            864 S E14+NT
L54
L55
              2 S L54 AND L51
L56
              10 S L51 AND (BREAKPOINT OR BREAK POINT) NOT L53, L55
L57
             367 S L51 AND SIZE
L58
               2 S L57 AND L10
L59
               1 S L58 AND GENOMIC LIBRARY/CT
L60
              10 S L44, L59 AND L1-L59
L61
              8 S L60 AND CHROMOSOM?
L62
              10 S L60 AND (DNA? OR GENOM?)
L63
              8 S L60 AND CLON?
L64
              10 S L60-L63
     FILE 'MEDLINE' ENTERED AT 16:21:59 ON 15 JUL 2003
     FILE 'BIOSIS' ENTERED AT 16:23:53 ON 15 JUL 2003
                E COLLINS C/AU
            754 S E3-E26,E80-E82
L65
                E VOLIK S/AU
L66
             16 S E3-E7
                E GRAY J/AU
L67
            699 S E3, E40
                E GRAY JOE/AU
L68
            140 S E3, E5
              2 S E30
L69
           1536 S L65-L69
L70
              1 S L70 AND L6, L5, L10
L71
L72
            695 S L70 AND (CONFERENCE/DT OR 00520/CC)
L73
            787 S L70 AND (CONFERENCE OR CONGRESS? OR POSTER OR SYMPOS? OR MEET
L74
             93 S L73 NOT L72
L75
             67 S L74 NOT ARTICLE/DT
L76
            694 S L72 AND L73
L77
            695 S L72, L76
L78
            309 S (10052 OR 10062 OR 10054 OR 10064)/CC AND L77
             25 S L77 AND 04500/CC
             21 S L77 AND 00530/CC
              9 S L78 AND L79, L80
            130 S 03508/CC AND L78
              7 S L82 AND L79, L80
              7 S L81 AND L83
              2 S L81 NOT L84
              8 S L71, L84
                SEL DN AN 5-8
L87
              4 S L86 NOT E1-E8
T88
              4 S L87 AND (10052 OR 10062 OR 10054 OR 10064 OR 03504)/CC
     FILE 'BIOSIS' ENTERED AT 16:41:30 ON 15 JUL 2003
L89
             29 S L79, L80 NOT L81, L83-L88
                SEL DN AN L89 12
L90
              1 S L89 AND E9-E10
L91
            177 S L78 NOT L79-L90
                SEL DN AN 5 7-11 31 40 50
L92
              9 S L91 AND E11-E28
L93
            357 S L77 NOT L78-L92
                SEL DN AN 3 6 30 31 39 44 60 61 60 70
              9 S L93 AND E29-E46
L94
L95
             23 S L93 AND COMPAR?(L)GENOM?(L)HYBRID?
L96
             7 S L94 AND L95
L97
             16 S L95 NOT L96
L98
             25 S L94-L97 NOT L88, L90, L92
```

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FILE 'HCAPLUS' ENTERED AT 16:53:17 ON 15 JUL 2003
                 E COLLINS C/AU
L99
             611 S E3-E27,E126-E132
                 E VOLIK S/AU
              22 S E3-E7
L100
                 E GRAY J/AU
L101
             205 S E3, E42, E43
                 E GRAY JOE/AU
             159 S E3, E5, E6
L102
L103
               2 S E65
             951 S L99-L103
L104
              64 S END SEQUENCE PROFIL?
L105
L106
               4 S L104 AND L105
L107
             104 S END? SEQUENC? PROFIL?
· L108
               4 S L104 AND L107
L109
             4 S L106, L108
                 SET HIGH ON
L110
              4 S L109 AND L99-L109
L111
              38 S L99 AND L100-L103
L112
              10 S L100 AND L101-L103
L113
              10 S L111 AND L112
L114
              10 S L110, L113
L115
              28 S L111 NOT L114
                 SEL DN AN 3 7 9 10 14 15 25 27 28
L116
               9 S L115 AND E1-E27
L117
              19 S L114, L116
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FILE 'HCAPLUS' ENTERED AT 17:02:04 ON 15 JUL 2003